Fundação Oswaldo Cruz
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Manifestações orais e dentárias em pacientes com Deficiências de Fagócitos: Uma Revisão Sistemática da literatura científica

Ana Luiza Machado Pinto

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Dissertação apresentada à Pós-Graduação em Saúde da Criança e da Mulher como parte dos requisitos para a obtenção do título de Mestre

Orientadora: Maria Ignez Capella Gaspar Elsas
Co-orientador: José Marcos Telles da Cunha

Junho, 2011

FICHA CATALOGRÁFICA NA FONTE
INSTITUTO DE COMUNICAÇÃO E INFORMAÇÃO
CIENTÍFICA E TECNOLÓGICA EM SAÚDE
BIBLIOTECA DA SAÚDE DA MULHER E DA CRIANÇA

P659m Pinto, Ana Luiza Machado
Manifestações orais e dentárias em pacientes com deficiências de fagócitos: uma revisão sistemática da literatura científica./ Ana Luiza Machado Pinto, 2011.
126 f. ; il.; tab.

Dissertação (Mestrado em Saúde da Criança e da Mulher) – Instituto Fernandes Figueira, Rio de Janeiro, R J, 2011.

Orientador: Maria Ignez Capella Gaspar Elsas
Coorinetador: José Marcos Telles da Cunha

Bibliografia: f. 123-126.

1. Anormalidades congênitas. 2. Fagócitos. 3. Imunodeficiência. 4. Manifestações bucais. 5. Infecções. I.Título.

CDD - 22ª ed. 610.043

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AOS MEUS QUERIDOS PAIS

ALBERTO E SILVINA,

Isto não seria possível sem vocês...

Obrigada pela vida, pelos ensinamentos e pelo amor incondicional. Obrigada por acreditarem sempre em mim e por me apoiarem em cada uma das minhas decisões.

Dedico esta conquista a vocês, meus amados pais!

Obrigada por tudo. Amo muito vocês!

A minha irmã Laura,

Obrigada pelo seu eterno incentivo, pela sua torcida e pelo seu carinho...

Amo muito você!

Ao meu amado marido Giovanni,

Obrigada pelo amor, carinho e companheirismo... Obrigada por acreditar em mim, por estar ao meu lado nos momentos mais difíceis, por compreender as minhas ausências e por me fazer assim, simplesmente muito feliz...

Eu te amo!

AGRADECIMENTOS

A **Deus**, pois sempre guiou os meus passos em todas as oportunidades que me proporcionou...

A minha querida orientadora **Maria Ignez Capella Gaspar Elsas**, por toda a paciência, dedicação, orientação e incentivo durante esses dois anos de mestrado. Muito obrigada por ter ficado sempre ao meu lado e ajudado a superar cada um dos obstáculos, pelos ensinamentos, por ter vibrado com cada vitória, mas também por todas as críticas que com certeza também foram importantes para a minha formação. O meu eterno agradecimento e admiração. Muito obrigada!

Ao meu querido co-orientador, **José Marcos Telles da Cunha**, por todos os seus ensinamentos e carinho. Obrigada por ter sido muito importante para a minha chegada até essa instituição e por sempre torcer pelo meu sucesso.

A minha querida família, dinda **Maria**, dindo **Albino** e avó **Laura** por torcerem muito pelo meu sucesso, por todo amor e carinho de sempre.

As minhas queridas amigas de coração e de profissão, **Ana e Dani**. Amigas que eu conquistei na faculdade e vão ficar para a vida toda. Obrigada por tudo que vivemos juntas e por estarem sempre ao meu lado, por me ouvirem, por me divertirem, por vibrarem com as minhas vitórias e por nunca terem me deixado desanimar. Eu amo muito vocês!

A todas as minhas queridas amigas: **Chris Belinho, Márcia Valéria** e **Cris Menezes**, pela amizade incondicional... Obrigada por estarem sempre ao meu lado, me aturando e torcendo pelas minhas vitórias. Amo vocês!

A **CAPES** pela bolsa de estudo concedida durante o curso de mestrado.

A todos que, direta ou indiretamente, contribuíram para a realização desta dissertação, o meu sincero agradecimento!

Resumo:

Os fagócitos são elementos fundamentais na resposta imunológica a diversos patógenos. Nas imunodeficiências primárias, defeitos quantitativos ou qualitativos do desenvolvimento do sistema imune, que afetam esta classe de células são responsáveis por um aumento do número de infecções graves ainda na primeira infância, que pode acometer, entre outros sítios, a cavidade oral destes pacientes.

Apesar da grande importância clínica, pela gravidade das manifestações e pela cronicidade da doença, as imunodeficiências primárias apresentam dificuldades importantes para o não-especialista, pela sua relativa raridade na população, pela grande heterogeneidade de mecanismos patogênicos, e pela diversidade de apresentações clínicas. Assim, a freqüência e a natureza das manifestações na cavidade oral dependem da natureza do defeito na imunidade, podendo variar consideravelmente de uma doença a outra. No entanto, um estudo detalhado da literatura científica sobre manifestações orais nas diferentes categorias de imunodeficiência primária. Neste trabalho, procuramos suprir parcialmente esta lacuna, fazendo uma revisão sistemática dos relatos de casos das imunodeficiências de células fagocitárias, e estabelecendo a freqüência das manifestações orais e dentárias descrita para esta classe de doenças, com o intuito de aprimorar o olhar do odontólogo na abordagem destes pacientes.

Resultados: A presente revisão sistemática do nosso estudo permitiu a avaliação de 1721 pacientes e entre estes pacientes, 653 pacientes (37,9%) apresentaram relato de alguma manifestação oral e/ou dentária na descrição clínica do caso. A doença periodontal foi a manifestação oral mais freqüente

(54,5%), seguida da perda precoce de dentes decíduos, encontrada em 142 pacientes (21,7%) e da gengivite, encontrada em 72 pacientes (11,0%), além da presença de aftas (8,1%) e a candidíase oral (7,5%)

Palavras-chave: imunodeficiência primária, defeitos congênitos de fagócitos, infecções, manifestações orais

Abstract:

Phagocytic cells are essential elements in the host reponse to a wide variety of pathogens. In Primary Immune Deficiency (PID) diseases, quantitative or qualitative defects in the development of the immune system, affecting phagocytes, account for an increase in the number of severe infections in infancy and childhood, which may involve, among other sites, the oral cavity.

Despite their great clinical relevance, in view of the diverse manifestations and chronicity of the disease, PID present important practical difficulties for the nonspecialist practicioner, due to their relative scarcity in the general population, great heterogeneity in pathogenetic mechanisms, and diversity of presentation. Therefore, the frequency and nature of oral manifestations depend on the nature of the defect in immunity, varying considerably among specific PID. To our knowledge, there is no systematic study of the existing scientific literature with respect to oral manifestations in different subtypes of PID. In this study, we attempted to fill this gap, by carrying out a systematic review of case reports of PID affecting phagocytes, and establishing the frequency of the different oral and dental manifestations in this group of PID, with the goal of providing dental health professionals with more accurate information concerning these patients.

Results: Case reports describing 1721 patients enabled us to detect reports of oral or dental manifestations in the clinical description of 653 pacients (37,9%). Periodontal disease was the most frequent oral manifestation (54,5%), followed by the early loss of decidual teeth, which was found in 142 pacients (21,7%) and gingivitis, found in 72 pacients (11,0%). Ulcerations (8,1%) and oral candidiasis (7,5%) were also reported. This analysis also provided evidence that recent advances in biomedical research, with an increasing focus

on molecular analyses, significantly influenced the content of case reports, which are nowadays more often focused on the identification of mutated genes, rather than on the detailed description of clinical findings. As a result, online and computer-assisted information retrieval strategies do not necessarily recover the same references, when articles are searched on the basis of clinical descriptions, or on the basis of well-characterized molecular defects.

Keywords: primary immune deficiency, congenital phagocyte defects, infection, oral manifestations.

1) Introdução

Antes do advento da antibioticoterapia, é provável que muitos indivíduos com defeitos congênitos afetando o desenvolvimento ou a função do sistema imune morressem nos primeiros meses ou anos de vida, devido à susceptibilidade exagerada a determinados patógenos. Porém, estas mortes não eram corretamente atribuídas a uma doença imunológica, já que muitas crianças saudáveis também morriam de infecção (Janeway *et al*, 2007).

Na infância, o sistema imune encontra os antígenos ambientais pela primeira vez, configurando diferentes padrões de resposta imune (tolerância, imunidade ou hipersensibilidade) e/ou adquirindo a memória imunológica para aqueles que efetivamente induziram sensibilização. As crianças, em contato com outras crianças na família ou na creche, estão expostas a muitos patógenos, sendo, portanto, vulneráveis a infecções. A infecção, recorrente ou persistente, é o principal sinal sugestivo de imunodeficiência primária (IDP), e o maior motivo de procura por uma avaliação médica. As infecções em portadores de IDP também podem ser devidas a microorganismos oportunistas, com uma evolução clínica atípica. Ainda que muitas crianças com infecção de repetição apresentem um sistema imune normal, é importante reconhecer aquelas crianças que possam apresentar um quadro de IDP, investigá-las e, se confirmado o diagnóstico, tratá-las precocemente e de forma adequada (Janeway *et al*, 2007; Stiehm *et al*, 2004)

As IDPs compõem um amplo grupo de desordens genéticas heterogêneas, que afetam tanto os componentes da imunidade inata (como os neutrófilos, macrófagos, células dendríticas, células NK, proteínas do complemento), como os da imunidade adaptativa (como os linfócitos B e T)

(Geha *et al*, 2007). Trata-se de uma classe de doenças monogênicas, com padrão de herança variável, podendo ser ligado ao cromossomo X, ou autossômico (recessivo ou, mais raramente, dominante), ou ainda ser resultante de novas mutações (Bonilla *et al*, 2003). Em alguns casos, estados de IDP não seguem um único padrão de herança, embora sejam claramente influenciados por fatores genéticos (Stiehm *et al*, 2004), o que indica efeitos compartilhados por múltiplas mutações distintas.

Estes defeitos de desenvolvimento do sistema imune e de maturação da resposta imunológica acarretam manifestações clínicas bastante distintas, de acordo com o compartimento afetado. Tais diferenças na apresentação clínica servem de base para o diagnóstico clínico inicial, orientando a avaliação progressiva da função imunológica destes pacientes (Janeway *et al*, 2007)

As manifestações clínicas mais comuns, mas não as únicas, das imunodeficiências são as infecções. As IDPs, na infância, geralmente apresentam-se com infecções incomuns, recorrentes e graves, ou infecções causadas por microorganismos de baixa virulência, assim como alterações do sistema imunológico associadas a síndromes genéticas (Stiehm *et al*, 2004)

O odontólogo é um dos profissionais da área de saúde que tem maior oportunidade de detectar, numa fase relativamente precoce, as manifestações clínicas da IDP que acometem a cavidade oral, e, conseqüentemente, orientar os pacientes e suas famílias a buscar uma investigação mais aprofundada da doença. Isto se explica pela freqüência com que manifestações de infecção e alterações na cavidade oral se apresentam nos quadros de IDP. Desta forma, é de interesse estabelecer até que ponto estas manifestações são constantes na

literatura especializada e de que forma a informação científica a respeito pode contribuir para uma melhor capacitação desses profissionais na sua prática.

Este trabalho traz uma análise sistemática da literatura científica sobre as manifestações orais e dentárias associadas a uma categoria específica de IDP, os defeitos congênitos do número e ou função dos fagócitos (tabela V da Classificação de Imunodeficiência Primária) (Notarangelo *et al*, 2009) **(Anexo 1)**. Ao fazer esta análise, buscamos identificar as alterações orais e dentárias mencionadas nos relatos de casos de pacientes portadores das diferentes doenças causadas pela alteração de fagócitos.

Os fagócitos possuem um papel fundamental na defesa contra bactérias e fungos (Notarangelo *et al*, 2009). Os defeitos quantitativos e qualitativos das células fagocitárias resultam em infecções graves e recorrentes de origem bacteriana e fúngica (principalmente por *Candida* e *Aspergillus*), localizadas no trato respiratório, tecido subcutâneo, pele, membranas mucosas e linfonodos. (Notarangelo *et al*, 2009; Szczawinska-Poplonik *et al*, 2009).

As alterações intra-orais e dentárias são achados frequentes nos pacientes com deficiência de número ou função de células fagocitárias. A gengivite, a doença periodontal, as infecções orais de repetição (candidíase e herpes), úlceras aftosas e hipoplasia dentária são algumas destas alterações. Estas alterações podem indicar um quadro de imunodeficiência primária a ser investigado, assim como podem também colaborar para a confirmação de um diagnóstico. (Szczawinska-Poplonyk *et al*., 2009).

A literatura brasileira carece de uma referência acessível e atualizada sobre as possíveis alterações orais e dentárias, que podem ser encontradas durante consultas odontológicas de rotina, e que devem ser entendidas como

sinais de alerta para IDPs, inclusive para as doenças de fagócitos. Nesta situação, a importância do olhar clínico do cirurgião-dentista deve vir respaldada por informação confiável, sumarizada a partir de estudos originais. Como estas doenças costumam evoluir desfavoravelmente, se não forem diagnosticadas numa fase precoce, este trabalho tem por objetivo fornecer aos colegas envolvidos na assistência odontológica, informações científicas de grande interesse para sua prática.

2) Justificativa

Existem muitos desafios para os profissionais que se especializam na pesquisa, educação e cuidados clínicos e terapêuticos dos pacientes com IDPs. O primeiro destes desafios é acompanhar o rápido desenvolvimento da genética, na tentativa de reconhecer os defeitos moleculares envolvidos nestas doenças. O segundo desafio é traduzir estas descobertas genéticas e moleculares em novas terapias eficazes para os pacientes com imunodeficiência. O terceiro e, provavelmente, o maior desafio é convencer o sistema público de saúde da necessidade de financiar mais pesquisas e da importância de mais educação e de maior suporte a estes pacientes. (Shearer, 2007)

As IDPs são desordens intrínsecas que afetam o sistema imunológico em graus variados de gravidade. Estas doenças, por serem raras e complexas, acarretando um consumo de recursos diagnósticos e terapêuticos desproporcional ao número de indivíduos afetados, podem ser consideradas uma questão importante para o gerenciamento de custos para a saúde pública.

Numa fração importante de casos, as IDPs não são corretamente, ou precocemente, diagnosticadas e, uma grande parcela dos pacientes acometidos passa por múltiplas infecções e internações antes de receber um diagnóstico adequado. O diagnóstico adequado e precoce é fundamental para a redução da morbimortalidade destas doenças.

Neste sentido, diversos países, já reconheceram o impacto destas desordens na população, o que permitiu a organização de políticas públicas voltadas para a abordagem e o acompanhamento destes pacientes. (Lindegren *et al*, 2004). No Brasil, no entanto, as IDPs ainda não conquistaram seu espaço entre as políticas de saúde pública.

As IDPs estão associadas a diversas alterações que acometem os tecidos moles e duros da cavidade oral, bem como a forma, estrutura e o número de dentes. Em proporção significativa de casos, estas alterações podem indicar um possível quadro de IDP a ser investigado. Por exemplo, a doença periodontal agressiva, quando presente em crianças, pode sugerir uma possível deficiência de fagócitos. As manifestações intra-orais e dentárias formam, portanto, um conjunto de alterações importantes, frequentemente encontradas nas Imunodeficiências Primárias. (Szczawinska-Poplonyk *et al*, 2009).

Durante as últimas décadas, muitos estudos voltaram-se para as manifestações orais da infecção pelo vírus da imunodeficiência humana (HIV). Certas alterações encontradas na Síndrome de Imunodeficiência Adquirida, tais como a candidíase oral, as úlceras orais e a doença periodontal, também podem ser encontradas nos pacientes com IDP. Porém, ainda que estas

manifestações intra-orais e dentárias sejam freqüentes nestes últimos, existem poucos estudos nesta área.

Durante o período em que acompanhei, como voluntária, o Ambulatório de Imunodeficiência Primária do IPPMG, pude observar a dificuldade no acompanhamento dos pacientes com suspeita de um possível quadro de IDP, bem como a angústia de sua famílias. Os pacientes costumam chegar ao serviço já numa fase avançada da evolução da doença, sem um diagnóstico preciso, já com seqüelas de difícil tratamento e, muitas vezes, sem a possibilidade de acesso a todos os exames necessários.

Diante da necessidade de entendermos a epidemiologia dos achados das manifestações orais e dentárias em IDP, optamos por realizar um estudo de revisão sistemática das imunodeficiências de fagócitos - Tabela V da Classificação de Imunodeficiência Primária (Notarangelo, 2009), entendendo o papel-chave destas células na resposta imunológica a infecções orais, sobretudo na doença periodontal.

3) Objetivos:

3.1) Geral

Realizar uma revisão sistemática da literatura científica, para identificar as manifestações orais e dentárias nos relatos de casos de pacientes com defeitos congênitos de número e/ou função de fagócitos.

3.2) Específico:

- Determinar a freqüência de manifestações orais e dentárias descritas em relatos de casos da literatura científica de pacientes com defeitos congênitos de número e/ou função de fagócitos.

- Definir as freqüências das diferentes manifestações orais e dentárias relatadas para cada um dos principais defeitos congênitos de número e/ou função de fagócitos.

4) Quadro teórico

4.1) Conceito e histórico

As imunodeficiências primárias (IDPs) formam um grupo de doenças que, devido a uma ou mais anormalidades do sistema imunológico, aumentam a susceptibilidade a infecções. (Stiehm *et al*, 2004)

Antes do advento da antibioticoterapia, é provável que muitos indivíduos com defeitos congênitos da imunidade morressem nos primeiros meses ou anos de vida, devido à susceptibilidade exacerbada a determinados patógenos. Porém, estas mortes não eram atribuídas a uma debilidade específica do sistema imune, já que muitas crianças saudáveis também morriam de infecção

(Janeway, 2007). Apenas após a Medicina ter possibilitado a cura de infecções graves como pneumonia, meningite, celulite e outras, é que estas doenças puderam ser melhor identificadas (Stiehm *et al*, 2004).

Mesmo assim, algumas síndromes de imunodeficiência como a candidíase crônica mucocutânea (Thorpe e Handle, 1929), a ataxia-telangiectasia (Syllaba e Henner, 1926) e a Síndrome de Wiskott-Aldrich (Wiskott, 1937), foram descritas antes mesmo de 1940 (Stiehm *et al*, 2004). Após esta década, foi descrito um caso de um paciente com defeito de imunidade celular por Glanzmann e Riniker (1950) e um caso de agamaglobulinemia em uma criança suíça por Hitzig (1958).

A primeira doença de IDP propriamente dita, ou seja, independentemente de acometimento primário de outros sistemas, foi descrita somente em 1952 por Ogden C. Bruton, que relatou o caso de um menino que não produzia anticorpos, que mais tarde foi denominada de Agamaglobulinemia ligada ao X de Bruton. Desde então, já foram caracterizadas mais de 130 doenças que afetam o desenvolvimento e/ou a função do sistema imunológico (Janeweay *et al*, 2004; Notarangelo, 2010)

Estes defeitos de desenvolvimento do sistema imune e/ou de maturação da resposta imunológica acarretam manifestações clínicas bastante distintas, de acordo com o compartimento afetado; contudo, esta heterogeneidade não impede a realização de um diagnóstico clínico inicial, e, pelo contrário, fundamenta e orienta a avaliação progressiva da função imunológica destes pacientes. Por exemplo, infecções de repetição por bactérias extracelulares sugerem deficiência predominantemente de anticorpos (deficiências humorais), ou deficiência de proteínas do sistema complemento. Por outro lado, infecções

por bactérias intracelulares e microorganismos oportunistas sugerem deficiência da imunidade celular. Nas deficiências de fagócitos é comum a história de abscessos de repetição. As deficiências predominantemente humorais representam cerca de 60% das imunodeficiências primárias, seguida das deficiências celulares e de fagócitos, responsáveis por cerca de 20 e 18%, respectivamente. As deficiências de sistema complemento são as mais raras, somando cerca de 1% dos casos de IDP, na maior parte das séries de pacientes encontradas na literatura. (Bonilla *et al.* 2005).

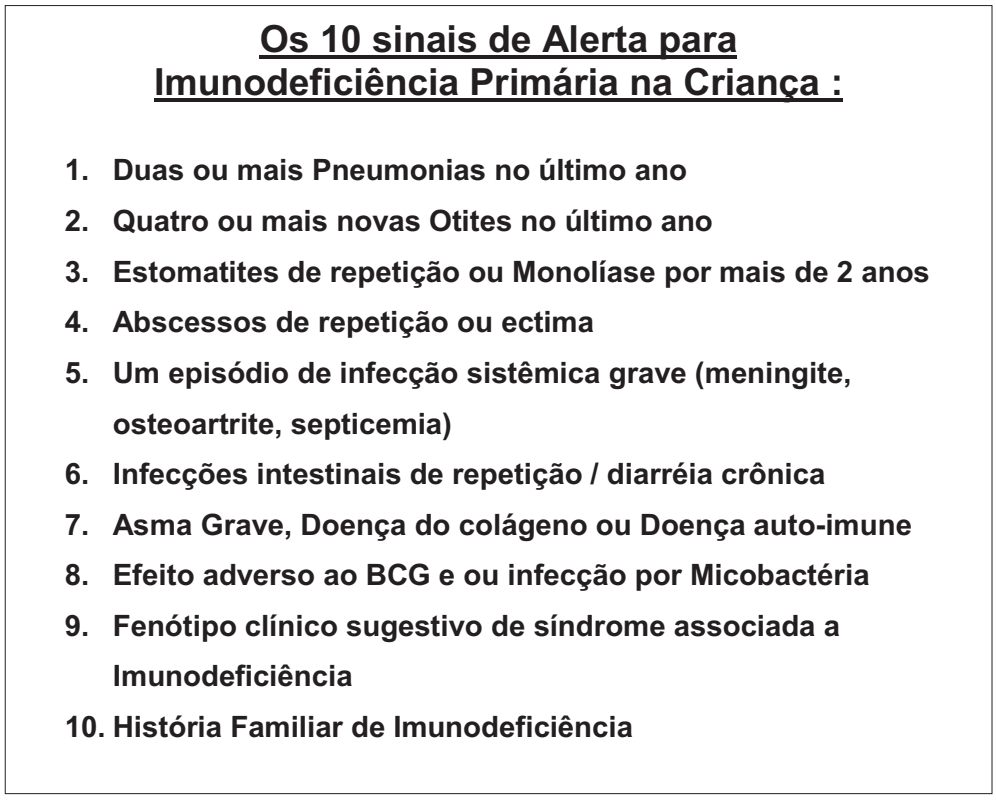
O diagnóstico precoce das IDPs é a chave para a sobrevivência desses pacientes, pois permite uma redução, tanto das complicações inerentes às múltiplas infecções e internações, quanto da mortalidade associada a estas doenças, além de permitir, nos casos com indicação, a realização de transplante de células-progenitoras hematopoiéticas (Buckley, 2006). O período entre o início dos sintomas e o diagnóstico de IDP é decisivo: quanto mais precoce for o diagnóstico, maior a chance de sobrevida destes pacientes, sobretudo naqueles pacientes portadores de Imunodeficiência Combinada Grave (SCID) (Muller e Friedrich, 2005; Elder *et al*, 2000).

Atualmente, temos no transplante de células-tronco hematopoiéticas (TCTH) e na terapia gênica (Chinen e Puck, 2004; Qasim *et al*, 2009) opções de tratamentos curativos, sobretudo para os casos mais graves, nos quais as reposições de imunoglobulina e a antibioticoprofilaxia não são suficientes para manter o paciente livre de infecções e de suas complicações.

A Fundação Jeffrey Modell, em colaboração com a Cruz Vermelha Americana, identificou e divulgou os “10 sinais de alerta” das IDPs (http: //www.jmfworld.org). A presença de mais de um desses sinais aponta para a

investigação mais apurada do paciente em questão. Esses critérios foram adaptados para a população brasileira pela Associação Brasileira de Alergia e Imunopatologia (ASBAI), pela Sociedade Brasileira de Pediatria (SBP) e pelo Grupo Brasileiro de Estudos de Imunodeficiências Primárias (BRAGID). (http://www.imunopediatria.org.br). Este documento conjunto tem sido divulgado essencialmente junto à classe médica, porém existem poucas informações divulgadas a respeito do alcance desta iniciativa. (**figura 1**)

Figura 1: Sinais de alerta para a investigação de IDP



(http://www.jmfworld.org. sd)

4.2) Classificação

Por conta do reconhecimento acelerado de novos defeitos genéticos, desde 1970, um comitê de especialistas se reúne a cada dois anos para atualizar a classificação das IDP, baseada nos principais mecanismos imunológicos implicados (Notarangelo *et al*, 2009). A mais recente atualização **(Anexo 1)**, publicada em dezembro de 2009, classifica as IDP em oito grupos, de acordo com o componente do sistema imunológico primariamente envolvido: a) imunodeficiências combinadas de células T e B (tabela I), b) deficiências predominantemente de anticorpos (tabela II), c) outras síndromes bem definidas (tabela III), d) doenças de desregulação imune (tabela IV), e) defeitos congênitos do número e/ou função de fagócitos (tabela V), f) defeitos na imunidade inata (tabela VI), g) doenças autoinflamatórias (tabela VII), h) deficiências de complemento (tabela VIII) (Notarangelo *et al*, 2009)

4.2.1) Imunodeficiências combinadas de células T e B

As Imunodeficiências Combinadas constituem um grupo heterogêneo de doenças, caracterizadas pela falha na imunidade mediada por células T e com comprometimento da função das células B, podendo inclusive acometer as células NK e mesmo a série mielóide. Este grupo de doenças estão agrupadas na tabela I da classificação de IDP (Notarangelo, 2009) **(anexo 1)**, onde estão contidas as formas mais graves de IDP, devido ao importante papel das células T na indução e modulação das respostas imunes específicas. (Stiehm *et al*, 2004)

Este grupo de doenças possui um início precoce, ainda nos primeiros meses de vida, não raro, muitas vezes associado a um quadro de diarréia prolongada, pneumonia intersticial, candidíase persistente e falha ao prosperar.

A Imunodeficiência Combinada Grave (SCID), a principal representante deste grupo, é geralmente fatal ainda no primeiro ano de vida, se não for realizado transplante de células-progenitoras hematopoiéticas (Kumar, 2006)

4.2.2) Imunodeficiências predominantemente de Anticorpos

As imunodeficiências resultantes da deficiência humoral ou de anticorpos representam o grupo mais numeroso entre as IDPs, cujas diferentes doenças estão agrupadas na tabela II da classificação de IDP (Notarangelo, 2009) **(anexo 1)**. Estas doenças podem ser detectadas precocemente, entre os 6 e 12 meses de idade, devido à perda gradual da IgG de origem materna, ou podem ser identificadas mais tardiamente. Este grupo apresenta um espectro clínico bastante variável, que abrange desde pacientes assintomáticos até manifestações clínicas bastante graves. (Kumar, 2006)

A agamaglobulemia ligada ao X caracteriza-se por infecções recorrentes por bactérias encapsuladas, devido a uma interrupção na diferenciação dos linfócitos B, resultando em baixos níveis de todas as classes de imunoglobulinas ou ausência total de produção de anticorpos. (Stiehm *et al*, 2004)

A deficiência seletiva de IgA é a mais freqüente IDP, e é definida pela dosagem de IgA sérica menor que 7 mg/dl em crianças com 4 anos ou mais. A maioria dos pacientes com deficiência de IgA são assintomáticos, mas

apresentam uma maior incidência de infecções, especialmente sinopulmonares, doenças autoimunes, atopia e doença celíaca. (Young, 2008)

4.2.3) Outras síndromes de imunodeficiência bem definidas

É um grupo heterogêneo de síndromes clinicamente bem caracterizadas e com mapeamento genético definido, porém que não atendem aos critérios necessários para integrarem aos outros grupos de IDP, compondo as doenças da tabela III da classificação de IDP (Notarangelo, 2009) **(anexo 1)**.

Neste grupo destaca-se a Síndrome de Wiskott-Aldrich, que se caracteriza por trombocitopenia (com microplaquetas), eczema e imunodeficiência (infecções recorrentes e um risco aumentado de auto-imunidade e neoplasia) (Notarangelo, 2010). Esta síndrome apresenta um padrão de herança ligado ao cromossomo X e um fenótipo variado que se correlaciona com o tipo de mutação no gene que codifica a proteína da Síndrome de Wiskott –Aldrich (WASp). (Stiehm *et al*, 2004)

A Síndrome de Hiper IgE caracteriza-se pelo nível elevado de IgE associado a eczema e susceptibilidade a infecções cutâneas e pulmonares (com formação de pneumatoceles) por *S. aureus* e *Candida*. Possui padrão autossômico dominante e recessivo. A Síndrome de Hiper IgE autossômica dominante é resultado do defeito do gene STAT-3 e apresenta retenção de dentes decíduos, escoliose, risco aumentado de fraturas ósseas, hiperextensibilidade das articulações e aspecto facial característico. O padrão autossômico recessivo está associado a mutação do gene de Tyk-2 (Notarangelo, 2010).

4.2.4) Doenças de Desregulação Imunológica

Algumas formas de imunodeficiência agrupadas na tabela IV da classificação de IDP (Notarangelo, 2009) **(anexo 1)** caracterizam-se por manifestações auto-imunes significativas devido a um distúrbio de homeostase imunológica (Notarangelo, 2010).

A desregulação da homeostase dos linfócitos pode gerar três conseqüências principais: acúmulo anormal de linfócitos, autoimunidade pela falha de remoção de linfócitos autorreativos e aumento na ocorrência de linfomas pela sobrevivência inapropriada de linfócitos transformados (Stiehm *et al*, 2004).

A síndrome linfoproliferativa autoimune (ALPS) é uma representante deste grupo de doenças. Os sintomas surgem precocemente, por volta dos 24 meses de idade. A linfoadenomegalia e/ou esplenomegalia persistente, podendo ser acompanhado por hepatomegalia, sem evidências de doença hepática e aumento de volume do timo são alguns dos achados clínicos da ALPS. A maioria dos pacientes apresenta linfocitose T e B, eosinofilia e/ou monocitose. A presença de auto-anticorpos é freqüente, sobretudo os anti-cardiolipina. O perfil laboratorial típico é a presença de número aumentado de células T duplo negativos no sangue periférico, ou seja, linfócitos T periféricos que não expressam CD4 nem CD8 (Notarangelo, 2010).

4.2.5) Defeitos congênitos de número e/ou função dos fagócitos

Os pacientes com deficiência de número e/ou função de células fagocitárias sofrem de infecções recorrentes e graves por bactérias (*S. aureus*

e *Serratia*, principalmente) e fungos, predominantemente na pele e trato respiratório (pneumonia), abscessos profundos, estomatites orais de repetição, doença periodontal e osteomielite (Notarangelo, 2010; Fleisher *et al*, 2007).

Por ser o objeto de estudo na presente dissertação, este grupo, que corresponde às doenças agrupadas na tabela V da classificação de IDP (Notarangelo *et al*, 2009) **(anexo 1)** será abordado em um capítulo à parte.

4.2.6) Defeitos da imunidade inata

A imunidade inata é a primeira barreira de defesa imunológica encontrada pelos agentes infecciosos. Os seus mecanismos são rapidamente ativados na presença de algum patógeno, antes mesmo da ativação do sistema imune adaptativo. Os componentes da imunidade inata incluem as barreiras epiteliais, os neutrófilos, macrófagos e células NK, o sistema complemento e algumas citocinas (TNF, IL-1, IL-12 e IFN-gama). Os receptores da resposta imune inata reconhecem estruturas presentes em diversos tipos de agentes patogênicos, ausentes em mamíferos, conhecidas como PAMPs (padrões moleculares associados aos patógenos). Os principais receptores que reconhecem os PAMPs são *Toll-like-receptor* (TLR). (Stiehm *et al*, 2004)

Os defeitos da imunidade inata compõem a tabela VI da classificação de IDP (Notarangelo *et al*, 2009) **(anexo 1).**

4.2.7) Doenças autoinflamatórias

As doenças autoinflamatórias são caracterizadas por processos inflamatórios espontâneos, sem que haja envolvimento importante da imunidade inata (Stojanov e Kastner, 2005). Este grupo de doenças estão

agrupadas na tabela VII da classificação de IDP (Notarangelo *et al,* 2009) **(anexo 1)**

A Febre Familiar do Mediterrâneo é a doença mais comum e possivelmente a mais estudada dentro deste grupo de doenças autoinflamatórias. Os sintomas mais freqüentes são episódios de febre alta, acompanhados de peritonite, sinovite e pleurite. A dor abdominal pode ser confundida com apendicite aguda, principalmente quando associada a vômitos e diarréia. Os ataques ocorrem em intervalos irregulares e podem variar de ataques semanais até períodos de remissão de semanas a meses sem nenhuma explicação aparente (Padeh e Berkun, 2007).

4.2.8) Deficiências do Complemento

Uma variedade de defeitos intrínsecos do sistema complemento já foi relatada na literatura. As deficiências do sistema Complemento estão relacionadas na tabela VIII da classificação de IDP (Notarangelo, 2009) **(anexo 1).** A deficiência em um dos componentes da via clássica compromete a opsonização das bactérias, a atividade bactericida e a remoção dos imunocomplexos. As deficiências dos componentes iniciais da via clássica (C1q, C1rs, C1s, C2 e C4) causam manifestações semelhantes às encontradas no lúpus eritematoso sistêmico. Já as deficiências dos componentes finais da via do complemento (C5-C9) causam infecções recorrentes por *Neisseria* (Notarangelo, 2010).

As deficiências de lectina ligadora de manose (MBL), componente dependente da via MBL do complemento, estão associadas ao aumento do

risco de infecções bacterianas, especialmente durante os primeiros anos de vida (Notarangelo, 2010)

4.3) Defeitos congênitos de número e/ou função dos fagócitos

4.3.1) Papel dos fagócitos no sistema imunológico

A pele e as mucosas representam a primeira linha de defesa e atuam como uma barreira efetiva contra a maioria dos microorganismos. Os patógenos que conseguem atravessar as superfícies epiteliais e de mucosa, na maioria das vezes, são eficientemente removidos pelos mecanismos inatos que atuam nos tecidos.

Desta forma, as doenças infecciosas só ocorrem quando o microorganismo é capaz de “escapar” das defesas inatas do hospedeiro para, em seguida, estabelecer uma infecção e multiplicar-se (Janeway *et al*, 2007).

Se um patógeno atravessa a barreira epitelial e inicia o seu processo de replicação nos tecidos, ele poderá ser rapidamente reconhecido pelos fagócitos mononucleares, ou macrófagos, residentes nos tecidos. Estas células maturam a partir dos monócitos do sangue e deixam a circulação para migrar para os tecidos do organismo. Os macrófagos são numerosos em vários sítios, principalmente no tecido conjuntivo, na camada submucosa do trato gastrointestinal, nos alvéolos, nos sinusóides hepáticos (onde são conhecidos como células de Kupffer) e no baço, onde são responsáveis por remover as células senescentes do sangue. A segunda principal família de fagócitos são os leucócitos polimorfonucleares neutrófilos (PMN) ou, abreviadamente, neutrófilos. São células de vida curta, abundantes no sangue e recrutadas para o tecido somente no contexto da infecção (Janeway *et al*, 2007).

Os microorganismos que invadem os tecidos são reconhecidos pelos macrófagos, porém estes são rapidamente reforçados pelo recrutamento de um grande número de neutrófilos para o local da infecção. O reconhecimento dos patógenos pelos macrófagos e neutrófilos ocorre através de receptores de superfície que identificam moléculas de superfície expressa pelos patógenos (receptores para padrões moleculares associados a patógenos). A ligação a estes receptores ativa os mecanismos de fagocitose. A fagocitose é um processo no qual o patógeno ligado é envolvido pela membrana do fagócito e em seguida interiorizado dentro da vesícula assim formada, o fagossoma. Além disso, os macrófagos e neutrófilos possuem grânulos (enzimas, proteínas e peptídeos) circundados por uma membrana chamados de lisossomas, que podem mediar uma resposta intracelular antimicrobiana quando na fusão com o fagossoma para a destruição do patógeno. O fagossoma torna-se acidificado, após a sua fusão com os lisossomas, matando a maioria dos patógenos.

Além disso, os macrófagos e neutrófilos também produzem uma variedade de produtos tóxicos que ajudam a matar o microorganismo englobado, tais como o óxido nítrico, o ânion superóxido e o peróxido de hidrogênio (Janeway *et al*, 2007).

Os macrófagos e os neutrófilos são fundamentais na imunidade inata, pois reconhecem, ingerem e destroem vários tipos de patógenos, sem o auxílio da imunidade adaptativa, apresentando um papel crucial na defesa inicial contra bactérias e fungos (Notarangelo, 2010).

4.3.2) Imunodeficiências causadas por alterações em número e função de fagócitos

Os pacientes com deficiência de número e/ou função de células fagocitárias sofrem de infecções recorrentes e graves de origem fúngica e bacteriana, com predomínio das infecções cutâneas e de trato respiratório, abscessos profundos e estomatites de repetição (Notarangelo, 2010).

4.3.2.1) Neutropenias Congênitas

A neutropenia congênita grave é um termo clínico que descreve uma contagem de neutrófilos abaixo de 500/µl, independentemente da causa. Podem ser subdivididas em dois grupos: Neutropenia congênita grave associada ao defeito na maturação de células mielóides (Síndrome de Kostmann) e Neutropenia congênita grave com outras características (Bohn, 2007).

As neutropenias congênitas são imunodeficiências primárias raras, caracterizadas por infecções bacterianas graves de início precoce, e neutropenia persistente devido à deficiência quantitativa de neutrófilos. São doenças geneticamente heterogêneas, com diferentes padrões de herança (autossômica dominante, autossômica recessiva, ligada ao X ou esporádica). (Stiehm *et al*, 2005).

O defeito genético da maioria dos casos de neutropenias congênitas de herança dominante atinge a elastase 2 *(ELA-2)*. A elastase de neutrófilos é uma serina-protease expressa em neutrófilos e monócitos, com atividades antimicrobianas e inflamatórias. (Bohn, 2007)

As manifestações clínicas mais freqüentes nas neutropenias congênitas são abscessos superficiais, úlceras orais, infecções cutâneas, onfalite, pneumonia e otite média. Durante a evolução da doença, os pacientes sofrem com abscessos em diversos locais, manifestações mucocutâneas, infecções respiratórias e diarréia (Rezaei et al, 2008).

4.3.2.2) Doença de Kostmann

A Síndrome de Kostmann é uma forma autossômica recessiva de neutropenia congênita grave, caracterizada por neutropenia persistente e profunda, associada a um característico atraso de maturação celular na medula óssea (Carlsson *et al*, 2007). Esta doença foi primeiramente descrita por Kostmann, em 1956, em uma família com infecções bacterianas graves e neutropenia persistente, devido ao defeito na maturação do estágio promielócito-mielócito. As manifestações clínicas são precoces e se caracterizam pela presença de febre, estomatite, infecções cutâneas e abscesso perirretal (Stiehm *et al*, 2004).

A presença do defeito genético na proteína *HAX-1* (“gene de Kostmann”) ocasiona o quadro de neutropenia congênita, grave devido ao aumento na apoptose dos progenitores mielóides (Bohn *et al*, 2007).

Outro defeito genético raro também associado à esta síndrome é a mutação no gene GFI-1, que ocasiona atraso de maturação das células mielóides, escassez de neutrófilos maduros e infecções de repetição (Klein, 2009).

4.3.2.3) Neutropenia com malformações cardíacas e urogenitais

Este tipo de neutropenia está associada a um defeito genético na via da G6PC3. Os pacientes com deficiência de G6PC3 apresentam neutropenia congênita devido ao atraso da maturação das células mielóides e aumento de apoptose dos neutrófilos periféricos, além de desordens variadas de desenvolvimento que afetam o sistema cardiovascular e/ou urogenital.

De acordo com Boztug e colaboradores, a via G6PC3 é necessária para a viabilidade dos neutrófilos, ressaltando o importante papel da glicose na homeostase dos neutrófilos (Boztug *et a*l, 2009).

4.3.2.4) Doença de armazenamento de glicogênio tipo 1B

A doença de armazenamento de glicogênio tipo Ib (GSD-Ib) é uma doença autossômica recessiva, com uma incidência de 1 para 500.000. Predominantemente considerada uma desordem metabólica caracterizada por hipoglicemia, excesso de acúmulo de glicogênio no fígado e rim e perfil metabólico sérico anormal, GSD-Ib é também uma doença imunológica, caracterizada por neutropenia e disfunções mielóides. Embora a base genética da doença, mutações no gene do transportador de glicose-6-fosfato (G6PT ou gene SLC37A4), esteja bem estabelecida, apenas o componente metabólico da doença é entendido bioquimicamente. (Choua *et al*, 2010).

4.3.2.5) Neutropenia Cíclica

A neutropenia cíclica é uma imunodeficiência primária autossômica dominante ou esporádica, caracterizada pela ocorrência de quadros de neutropenia a cada 3 semanas, com duração de 3 a 6 dias, e associada com algumas mutações em ELA-2.

Os pacientes com neutropenia cíclica congênita são geralmente assintomáticos entre os períodos neutropênicos, mas durante o episódio de neutropenia apresentam infecções específicas, principalmente em cavidade oral e mucosas, tais como estomatite aftosa recorrente, úlceras orais, gengivite, doença periodontal, além de abscessos cutâneos recorrentes e ocasionalmente infecções piogênicas graves (Rezaei *et al*, 2004).

4.3.2.6) Neutropenia ligada ao X

Esta doença caracteriza-se pelo ganho de função da proteína WASP (proteína da Síndrome de Wiskott-Aldrich), ocasionando a perda do mecanismo de auto-inibição da proteína e, consequentemente um aumento da polimerização da actina. Além da neutropenia, os pacientes apresentam graus variados de linfopenia, redução da proliferação de neutrófilos e ausência de atividade fagocítica (Klein *et al*, 2009).

4.3.2.7) Deficiência de P14

A p14 é uma pequena proteína adaptadora endossomal que captura a proteína cinase ativada por mitógenos (MAP cinase), dirigindo-a para o compartimento endossomal. Os pacientes com deficiência de p14 apresentam uma síndrome de imunodeficiência congênita complexa, caracterizada por

defeitos de pigmentação, falha do crescimento, neutropenia congênita grave, atraso na degradação dos patógenos ingeridos, além de imunodeficiência combinada de células T e B. Clinicamente, a deficiência de p14 lembra outras síndromes que apresentam redução de pigmentação e imunodeficiência, como a síndrome de Hermansky-Pudlak tipo 2 (HPS2), a síndrome de Griscelli tipo 2 (GS2), e a síndrome de Chediák-Higashi (CHS) (Teis *et al*, 2002; Bohn et al, 2008).

4.3.2.8) Deficiência de adesão leucocitária

As deficiências de adesão leucocitária (LAD) são um grupo de doenças raras, de herança autossômica recessiva, ocasionada pelo defeito na adesão do neutrófilo e caracterizada por úlceras, retardo de cicatrização e infecções bacterianas recorrentes. O fenótipo dos pacientes é variável e reflete a quantidade de $ß_2$ integrinas expressa na superfície celular dos leucócitos. Os pacientes com menos de 1% de expressão evoluem com infecções graves e alto risco de morte. Já aqueles com expressão entre 1-10% evoluem com defeitos na mobilidade leucocitária, aderência e endocitose, ocasionando quadros de doença periodontal, infecções cutâneas e retardo na cicatrização. Os familiares heterozigotos dos pacientes afetados têm expressão de $ß_2$ integrinas em torno de 40-60% e são clinicamente normais (Stiehm *et al*, 2004).

Estas doenças classificam-se em LAD-1, LAD-2 e LAD-3, que resultam em defeitos funcionais semelhantes, apesar de se originarem em mutações de genes totalmente distintos.

A deficiência de adesão leucocitária do tipo 1 (LAD-1) é a forma mais comum dentre as deficiências de adesão leucocitária e é decorrente da

mutação no gene ITGB2, que é responsável por codificar a cadeia β da família das β2 integrinas, também chamada de CD18. A ausência ou a síntese anormal de CD18 impede a expressão adequada de CD11 (Notarangelo e Badolato, 2009).

O heterodímero CD11/CD18 é expresso na superfície dos leucócitos e participa de muitos processos de defesa contra patógenos, incluindo adesão leucocitária, migração transendotelial, fagocitose e atividade citotóxica mediada por células (Notarangelo e Badolato, 2009).

A forma grave da doença está associada à ausência completa de CD18 e, estes pacientes cursam com sintomas clínicos mais acentuados e, consequentemente apresentam um maior risco de vida quando comparados aos pacientes que possuem uma expressão residual de CD18, suficiente para permitir adesão mediada por β-2 integrinas (Notarangelo e Badolato, 2009; Gazit *et al*, 2010).

A LAD-2 é causada pela mutação no gene FUCT1, que codifica a proteína transportadora de fucose GDP. Os pacientes com esta doença apresentam episódios de infecções recorrentes nos primeiros anos de vida, dismorfismo, baixa estatura e retardo mental significativo. (Gazit *et al*, 2010).

A LAD-3 foi reconhecida recentemente e cursa com defeitos combinados de ativação das integrinas dos leucócitos e plaquetas, impedindo a migração e outras funções. Está associada com mutações no gene KINDLIN3. Os pacientes com LAD-3 apresentam infecções recorrentes graves, tendência a sangramento e leucocitose. (Notarangelo e Badolato, 2009; Gazit *et al,* 2010)

4.3.2.9) Deficiência de Rac-2

Deficiência de Rac-2 ou Síndrome de Imunodeficiência de neutrófilo manifesta-se por um grave defeito na migração de leucócitos. As proteínas *Rho GTPases* (incluindo Rho, Rac, Cdc42) atuam como controles moleculares em respostas celulares amplas, como a reorganização da actina do citoesqueleto, a transcrição gênica e a proliferação das células. Seus papéis têm sido implicados, cada vez mais, na ativação de processos de sinalização em leucócitos, incluindo a transdução de sinal mediada por integrinas, a sobrevida celular induzida por fatores de crescimento e o processo de proliferação. Em particular, a alteração funcional de Rac-2, uma Rho GTPase especifica de células hematopoiéticas, ocasiona uma disfunção mielóide grave (Gu e Williams, 2002; Razeai *et al*, 2008)

Os pacientes com deficiência de Rac-2 apresentam defeitos importantes na quimiotaxia de neutrófilos, bem como inibição da produção de superóxido, ocasionando abscessos periumbilicais e perirretais, atraso na queda do coto umbilical e formação precária de pus (Gu e Williams, 2002; Razeai *et al*, 2008)

4.3.2.10) Deficiência de β-actina

A Deficiência de β-actina é uma deficiência autossômica dominante da polimerização de actina dos neutrófilos, acarretando um defeito na migração dos leucócitos. Tal como em pacientes com LAD, não há formação de pus no local da infecção.

Os pacientes sofrem com infecções bacterianas e fúngicas recorrentes sem formação de pus, além de retardo mental e fotossensibilidade (Razeai *et al*, 2008).

4.3.2.11) Periodontite juvenil localizada

A periodontite juvenil localizada é uma forma de doença periodontal de progressão rápida, que afeta a dentição permanente durante o período puberal, resultando na perda de inserção de no mínimo 4 mm em pelo menos dois primeiros molares e incisivos permanentes, independentemente do acúmulo de cálculo subgengival e da presença de inflamação gengival (Passanezi *et al*, 2007)

O mecanismo fisiopatológico da doença permanece desconhecido, porém observa-se, nesta doença, uma perda da quimiotaxia dos neutrófilos devido a uma mutação no receptor quimiotático (FRP1). (Razeai *et al*, 2008)

4.3.2.12) Síndrome de Papillon-Lefrève

A Síndrome de Papillon-Lefrève foi primariamente descrita por Papillon e Lefrèvre em 1924. Trata-se de uma doença autossômica recessiva, caracterizada por hiperceratose palmoplantar difusa e doença periodontal agressiva, afetando as dentições decídua e permanente. A doença é causada por uma mutação pontual no gene da catepsina C, que ocasiona um defeito funcional dos neutrófilos. (Razeai *et al*, 2008; Dhanrajani, 2009)

O início da doença geralmente está associado à erupção dos dentes decíduos, seguido de um quadro de inflamação gengival e rápida destruição do tecido periodontal, acompanhado pela perda precoce dos dentes decíduos. Este processo se repete quando na dentição permanente (Dhanrajani, 2009).

4.3.2.13) Deficiência de grânulos específicos

A Deficiência de grânulos específicos dos neutrófilos (SGD) é uma doença congênita rara. Os pacientes com esta doença possuem neutrófilos com núcleos bilobados atípicos, falta de expressão de pelo menos uma proteína primária e de todas as proteínas secundárias e terciárias dos grânulos, defeitos na quimiotaxia dos neutrófilos e desregulação da atividade bactericida. A falta de muitos constituintes dos grânulos resulta numa diminuição significativa da atividade bactericida independente de oxigênio, além de uma diminuição na expressão de moléculas de adesão e de receptores quimiotáticos na superfície da célula. O defeito é causado por uma mutação em um fator de transcrição especializado essencial à mielopoiese (CEBP ou proteína CCAAT), que regula a síntese de proteínas num período crítico de diferenciação dos neutrófilos, que abrange a parte final da produção de grânulos primários e todo o período de tempo durante o qual os grânulos específicos e os seus componentes são produzidos (Gombart e Koeffler, 2002).

Os pacientes com esta deficiência apresentam infecções bacterianas graves, que cursam com lesões ulcerativas e necrose da pele e mucosas, assim como pneumonias recorrentes, frequentemente causadas por *Staphylococcus aureus* e/ou *Pseudomonas aeruginosa,* com deficiência na formação do pus. Na hematoscopia, a segmentação anormal dos granulócitos é patognomônica. A quimiotaxia é significativamente reduzida, e os grânulos específicos estão ausentes, como evidenciado em imagens de granulócitos obtidas por microscopia eletrônica de transmissão. O diagnóstico definitivo é feito pela análise de mutação do gene CEBPε (fator de transcrição mielóide). (Gombart e Koeffler, 2002; Razeai *et al*, 2008)

4.3.2.14) Síndrome de Shwachman-Diamond

A Síndrome de Shwachman-Diamond (SDS) é uma doença autossômica recessiva que inclui insuficiência pancreática exócrina grave, falência de medula óssea, anormalidades esqueléticas e baixa estatura, com alteração no gene SBDS. É considerada, após a fibrose cística, a segunda maior causa de insuficiência pancreática exócrina em crianças (Razaei *et al*, 2008)

Os pacientes com SDS estão em risco de complicações, muitas vezes fatais, como infecções graves, leucemia mielóide aguda e falência de medula óssea *(http://www.shwachman.org/* acessado em 14 janeiro 2011)

4.3.2.15) Doença Granulomatosa Crônica ligada ao X

A Doença Granulomatosa Crônica (DGC) é uma deficiência de função das células fagocitárias causada pelo defeito no complexo de NAPH oxidase. Este complexo induz a produção de componentes antimicrobianos e a ativação de enzimas líticas, resultando na morte intracelular de bactérias e fungos. Os pacientes com DGC apresentam infecções recorrentes e por vezes graves, que acometem a pele, além abscessos hepáticos e perirretal e pneumonias com susceptibilidade a infecções por micobactérias (Doffinger *et al*, 2005; Razaei *et al* 2008; Holland, 2010).

A forma de DGC ligada ao X é decorrente de um defeito no gene *CYBB* que codifica a *gp91-phox* e responsável por cerca de 65% dos casos da doença.

Em geral, os pacientes com DGC ligada ao X apresentam quadros de infecções graves ainda em idade precoce. (Holland, 2010)

4.3.2.16) Doença granulomatosa crônica autossômica

A Doença Granulomatosa Crônica é uma desordem geneticamente heterogênea caracterizada pela ocorrência de mutações nos genes estruturais do complexo NADPH oxidase (Razaei et al, 2008). Inicialmente, somente a forma ligada ao X foi descrita. A forma autossômica recessiva foi posteriormente reconhecida no ano de 1968, em meninas. A forma autossômica recessiva deve-se a mutações em um dos três genes: o gene para p22*phox* (CYBA), o gene para p47*phox* (NCF1) e o gene para p67*phox* (NCF2) (Razaei *et al*, 2008; Ross, 1996). As manifestações clínicas nos pacientes afetados são semelhantes às descritas na forma da doença ligada ao X.

4.3.2.17) Deficiência da cadeia β1 do receptor de IL-12 e IL-23

A Via do interferon-γ (IFN-γ) / interleucina-12 (IL-12) tem papel central em muitos aspectos básicos e clínicos das vias de transdução de sinal de citocinas. Esta via é uma peça crucial no sistema imunológico e fundamental para o controle de infecções por micobactérias (Watford *et al*, 2004; Picard e Casanova, 2004)

A IL-12 regula tanto a imunidade inata como a adaptativa. No contexto da infecção micobacteriana, a principal ação da IL-12 é induzir a produção do IFN-γ por diferentes populações celulares do sistema imune. Assim como a IL-12, a IL-23 induz a produção de IFN-γ pelas células T. (Watford *et al*, 2004; Picard e Casanova, 2004; Rezaei et al, 2008) Esta redundância de efeitos tem

como base molecular o compartilhamento de uma subunidade sinalizadora do receptor, a cadeia β1, que é idêntica entre os receptores de IL-2 e de IL-23.

A deficiência de sinalização por IL-12/IL-23, que resulta de mutações no gene correspondente, acarreta uma IDP caracterizada pela susceptibilidade aumentada a micobactérias e a Salmonella, o que se explica pela deficiência da imunidade mediada pelo IFN-□. (Picard e Casanova, 2004)

4.3.2.18) Deficiência de IL-12p40

Mutações em IL12p40 também aumentam a suscetibilidade genética às doenças micobacterianas (Picard and Casanova, 2004). Neste caso, o mecanismo é distinto do da deficiência em cadeia β1, visto se tratar de uma deficiência no ligante e não no receptor, e não afetar, portanto, a sinalização redundante por IL-23. Além de ser crucial para o controle de micobactérias, a via IFN--γ/IL-12 também está envolvida na patogênese de doenças auto-imunes, bem como no desenvolvimento e controle do crescimento tumoral. Correlações genótipo-fenótipo têm sido estabelecidas para determinados genes desta via, alguns dos quais têm implicações terapêuticas. (Picard e Casanova, 2004)

4.3.2.19) Deficiência do receptor 1 do IFN-γ

As mutações no gene do IFN-γ-R1 foram as primeiras a serem identificadas como causa da susceptibilidade aumentada à doença por micobactérias, associada a um padrão mendeliano de herança. (Picard e Casanova, 2004)

As mutações com padrões de herança tanto recessiva como dominante resultam em deficiência do receptor 1 do IFNγ (IFN-γR1). A maioria das deficiências recessivas do IFNγR1 acarreta a perda completa de expressão do IFN-γR1 na superfície da célula e consequentemente a perda da capacidade de resposta ao IFN-γ. Estes defeitos são encontrados na deficiência recessiva completa (RC) do IFN-γR1. A deficiência recessiva parcial do IFN-γR1 acarreta uma resposta reduzida ao IFN-γ. (Doffinger *et al*, 2005; Picard e Casanova, 2004)

Os pacientes com deficiência RC de IFN-□R1 têm um fenótipo bastante grave quando comparado aos pacientes com uma deficiência parcial (DP), incluindo o início mais precoce, o aumento do número e da gravidade das infecções e diminuição da sobrevida (Picard e Casanova, 2004).

4.3.2.20) Deficiência do receptor 2 do INF-γ

O receptor de IFN-γR2 pertence também à família de receptores de citocinas classe II e é responsável pela transdução de sinal. A sua organização genética se assemelha à do IFN-γR1. As mutações em IFNγR2 foram identificadas menos frequentemente do que aquelas em IFN-γR1 (Picard e Casanova, 2004).

Os pacientes com deficiência completa de receptor 1 e 2 do IFN-□□apresentam infecções disseminadas graves por micobactérias causadas pelo BCG ainda na primeira infância, além da dificuldade de formação de granulomas. Já os pacientes com deficiência parcial, apresentam quadros mais brandos e, geralmente paucibacilares (Doffinger *et al*, 2005).

4.3.2.21) Deficiência de STAT 1

A STAT1 é fundamental na transdução do sinal mediado pelo IFN. A sua deficiência pode ser completa ou parcial. Os pacientes com deficiência completa também apresentam maior susceptibilidade a infecções virais, diferentemente dos pacientes com deficiência parcial. (Picard e Casanova, 2004)

Clinicamente, os pacientes com deficiência de STAT-1 apresentam infecção disseminada ao BCG e susceptibilidade a infecções por micobactérias. (Doffinger *et al* Kumararatne, 2005, Picard e Casanova, 2004;).

4.3.2.22) Síndrome de Hiper IgE

As síndromes de Hiper-IgE (HIES) são IDPs raras, caracterizadas por níveis séricos elevados de IgE, dermatite e infecções recorrentes do pulmão. Existem duas formas bem caracterizadas de HIES: uma forma dominante, causada por mutações no gene STAT3, e uma forma recessiva, associada a mutações no gene da Tirosina cinase 2 (Tyk2). Essas duas síndromes possuem diferentes apresentações e evoluções clínicas, e os mecanismos patogenéticos não são os mesmos, embora haja o achado comum de uma elevação de IgE. A forma dominante é caracterizada por um predomínio de alterações não-imunológicas, incluindo alterações nos tecidos esqueléticos, nos tecidos conjuntivos e nos pulmões, além de infecções recorrentes e eczema. Em contraste, a forma recessiva apresenta principalmente importantes infecções virais e complicações neurológicas. (Freeman e Holland, 2008)

4.3.2.22.1) Sindrome Hiper IgE autossômica dominante. (deficiência de STAT3)

Essa desordem foi primeiramente descrita como síndrome de Jó por Davis *et al*, em 1966, com referência às lesões de pele características. A síndrome foi revisada por Buckley et al, em 1972, que acrescentou à caracterização da doença a presença de níveis extremamente elevados de IgE sérica, passando assim a ser diagnosticada com a presença da tríade clássica - eczema, infecções recorrentes do pulmão e IgE elevada, sendo expandida para incluir alterações nos tecidos esqueléticos e conjuntivo, cardíacas e cerebrais. Recentemente foi demonstrado que as mutações em STAT3 são responsáveis pela maioria, senão pela totalidade, dos casos de HIES com herança autossômica dominante, e estas mutações, que são altamente pleiotrópicas, permitem explicar a natureza multissistêmica da doença. (Grimbacher *et al*, 1999; Freeman *et al*, 2008)

4.3.2.22.2) Sindrome Hiper IgE autossômica recessiva (AR hyper-IgE)

A síndrome de Hiper IgE autossômica recessiva é consideravelmente mais rara do que a forma autossômica dominante de HIES. O quadro é clinicamente distinto, mais grave, e pode estar associado com auto-imunidade e vasculite. Os pacientes possuem níveis séricos elevados de IgE, dermatite atópica grave, infecções estafilocócicas da pele e do pulmão, e infecções virais na pele e mucosas (mas sem anormalidades esqueléticas e dentárias), também apresentando suscetibilidade a bactérias intracelulares. A mortalidade na infância é elevada, geralmente devido à sepse bacteriana ou vasculite do SNC. Foi detectado que nesses pacientes a alteração molecular é no gene da

Tirosina cinase 2 (Tyk2), uma molécula importante na transdução de sinal para a IL-12, IL-6 e interferon alfa. A função fagocítica possivelmente estaria prejudicada, nesta síndrome, por defeitos na produção de citocinas necessárias à ativação dos fagócitos em condições fisiológicas (Heimall *et al*, 2010; Freeman *et al*, 2008; Minegishi e Karasuyama, 2008)

4.3.2.23) Proteinose pulmonar alveolar

A proteinose alveolar pulmonar é uma síndrome rara, caracterizada pelo acúmulo de lipoproteínas surfactantes que resulta da incapacidade de degradação deste material pelos macrófagos alveolares, insuficiência respiratória e susceptibilidade a infecções. A doença ocorre devido à produção de auto-anticorpos (a forma autoimune é a mais comum) contra o fator estimulador de colônias (GM-CSF) ou pela mutação do gene CSF2RA. (Martinez-Moczygemba and Huston, 2010; Trapnell et al, 2009)

O GM-CSF é crítico para a diferenciação dos macrófagos e suas funções imunológicas e para a homeostase e defesa pulmonar (Martinez-Moczygemba e Huston, 2010; Trapnell et al, 2009)

A forma congênita da doença está associada à mutação do gene CSF2RA, que ocasiona a desregulação da sinalização via GM-CSF. Os pacientes com proteinose alveolar pulmonar apresentam insuficiência respiratória e em casos mais graves, falência respiratória, devido ao acúmulo de surfactantes no espaço alveolar (Martinez-Moczygemba e Huston, 2010; Trapnell et al, 2009)

4.3.3) Manifestações orais nas imunodeficiências primárias de fagócitos

As manifestações orais e dentárias formam um grupo heterogêneo de alterações comumente encontrado nas IDP. Algumas anomalias dentárias, algumas lesões orais e a doença periodontal na infância podem ser sinais indicativos de um quadro de imunodeficiência a ser investigado. (Szczawinska-Poplonik *et al*, 2009).

Estudos demonstram que dentre as lesões mais freqüentes na cavidade oral de crianças saudáveis, destacam-se a candidíase oral, as estomatites aftosas recorrentes, o herpes labial recorrentes e as alterações de desenvolvimento da língua (língua geográfica e fissuras) (Bessa *et al*, 2004; Crespo *et al*, 2005). Nas IDP, estas manifestações iniciam-se em idade bastante precoce, são acompanhadas por processo de necrose tecidual com ulceração e invasão bacteriana, além de envolverem, em grande parte a saúde do tecido periodontal, ocasionando gengivite agressiva e doença periodontal (Szczawinska-Poplonik *et al*, 2009).

Nas IDPs ocasionadas por alterações de número e/ou função de fagócitos, dentre as manifestações descritas na literatura, destacam-se, sobretudo, as doenças do periodonto (gengivite e periodontite), com envolvimento do tecido ósseo de suporte dentário e conseqüentemente a perda precoce de dentes decíduos e permanentes (Szczawinska-Poplonik *et al*, 2009)

Como a doença periodontal depende da resposta imunológica do hospedeiro, é compreensível que muitas IDPs, principalmente as alterações de número e/ou função de fagócitos, comprometam a integridade do periodonto. Este fato relaciona-se a alteração quantitativa ou qualitativa (função) das células fagocitárias.

A doença periodontal é rara em crianças e adolescentes saudáveis. Quando presente corresponde apenas a um quadro de inflamação gengival, associado à placa bacteriana. No entanto, muitas desordens sistêmicas aumentam a susceptibilidade do paciente para a doença periodontal e, por vezes, este envolvimento é mais rápido e agressivo. Embora seja bem estabelecido que a principal causa da doença periodontal é a placa bacteriana, esta relação direta torna-se controversa, naqueles casos em que a doença periodontal está associada a determinadas desordens sistêmicas. Sugere-se que a predisposição sistêmica acelera o processo de destruição causado pelos agentes bacterianos. Porém já se sabe que o desenvolvimento e a evolução da doença periodontal dependem da resposta imunológica do hospedeiro, da integridade dos tecidos, da imunidade humoral e celular e de fatores endócrinos e nutricionais (Schenkein, 2006; Lindhe *et al*, 2005)

O defeito em qualquer etapa de neutralização de patógenos pelas células fagocitárias está associado ao aumento do risco de doença periodontal. (Szczawinska-Poplonik, 2009). Estudos experimentais têm demonstrado o papel dos neutrófilos na patogênese da doença periodontal e na expressão de moléculas inflamatórias (Schenkein, 2006).

Além da doença periodontal, dentre as manifestações que frequentemente acometem a cavidade oral dos pacientes com IDP, destacam-se a candidíase, a estomatite herpética e infecções recorrentes por herpes e a estomatite aftosa recorrente (Szczawinska-Poplonik *et al*, 2009; Aktinson *et al*, 2000).

A candidíase é a infecção fúngica bucal mais comum e também pode ocorrer em crianças saudáveis. A doença pode variar de um quadro leve, com

envolvimento superficial da mucosa a um quadro mais disseminado em pacientes com comprometimento imunológico (Neville, 1995). Dentre as desordens de número e/ou função das células fagocitárias, a candidíase é comumente encontrada nos pacientes com Síndrome de Hiper IgE e nos pacientes com desordens quantitativas ou qualitativas de neutrófilos (neutropenias congênitas e cíclicas e nas deficiências de adesão leucocitária) (Aktinson, 2000).

Cabe lembrar que a candidíase oral pode ocorrer em resposta ao uso de antibioticoterapia (Crespo *et al*, 2005) que muitas vezes é realizada nos pacientes com IDP a fim de evitar as infecções de repetição.

A infecção pelo vírus herpes é relativamente comum na população geral. A infecção primária pelo vírus pode ser assintomática ou sintomática (gengivoestomatite herpética primária – lesões de início abrupto, dolorosas que geralmente acometem crianças entre 6 meses a cinco anos de idade), apresentando, posteriormente como lesões secundárias e recorrentes, localizadas, principalmente, no vermelhão dos lábios e pele adjacente.

De acordo com o estudo de Szczawinska-Poplonik e colaboradores (2009), as infecções por herpes não estão entre as manifestações orais mais comuns nas desordens de fagócitos.

5) Metodologia

Este estudo é uma revisão sistemática da literatura científica, realizada em base de dados eletrônica *Medline/Pubmed*, conduzida entre os meses de agosto de 2010 a abril de 2011.

A primeira busca realizada utilizou como descritores [MeSh] ***"Immunologic deficiency syndromes AND stomatognathic diseases"***, com

limite em estudos humanos, com o objetivo de se obter uma análise fidedigna das alterações orais em IDPs.

A escolha pelo descritor *"**stomatognathic diseases"*** deveu-se ao fato deste descritor ser o descritor mais antigo na base de dados do *pubmed* e por entendermos a necessidade de alcançar o maior número de artigos possíveis.

Uma vez que escolhemos como alvo deste presente estudo as deficiências de número e/ou função das células fagocitárias e, por desejarmos analisar os relatos de casos dos pacientes com deficiências de fagócitos, nossos descritores cruzaram cada uma das doenças de fagócitos listadas na Classificação de IDP (Notarangelo, 2009) **(Anexo 1)** com o termo [MeSh] ***"case reports"***. Também foram feitas buscas que correlacionavam a doença de fagócitos com o devido defeito genético listado na Classificação de IDP (Notarangelo, 2009) **(Anexo 1).**

A realização de outras buscas mais abertas foi considerada sempre que houve necessidade de se alcançar um número maior de artigos a serem analisados, ou mesmo quando desejamos estabelecer comparações entre diferentes estratégias de buscas. Todas as buscas realizadas, com os seus respectivos descritores estão apresentadas sob forma de apêndices desta dissertação.

Foram incluídos na análise todos os artigos de relatos clínicos de pacientes com diagnóstico comprovado da doença de fagócito em questão. Os artigos que não apresentaram diagnóstico definido, que não possuíam descrição detalhada do quadro clínico, que não possuíam acesso eletrônico disponível e que não fossem em língua inglesa, espanhola ou portuguesa foram excluídos da análise.

Uma análise inicial foi realizada com base no título do artigo e nos resumos de todos os artigos que preenchiam aos critérios de inclusão ou nos artigos que não permitiam clareza de que deveriam ou não ser excluídos.

Após a análise dos resumos, os artigos selecionados para a análise foram devidamente examinados.

5.1) Coleta de dados

Os dados coletados por consulta online foram digitados em planilhas eletrônicas do Excel®. A planilha apresenta as seguintes variáveis: Nome do artigo, Nome do autor, código de identificação PMID da base eletrônica do Pubmed/Medline, presença ou ausência do artigo em meio eletrônico, tipo do artigo (relato de caso: sim ou não), quantitativo de pacientes analisados, diagnóstico confirmado e descrição idêntica das manifestações orais e dentárias conforme o surgimento na análise.

Os artigos que foram incluídos na primeira análise e que durante a leitura detalhada do texto não cumpriram os critérios de inclusão foram excluídos neste momento.

6) Resultados

6.1) Evolução da estratégia de busca:

A primeiro busca geral realizada com os descritores ***"immunologic deficiency syndromes AND stomatognathic diseases"***, limitando a estudos em humanos permitiu, além de estudos de imunodeficiência primária, uma grande quantidade de artigos relativos a HIV, HTLV e câncer, totalizando 4270 artigos. Esta forma de busca revelou-se ineficiente, porque alguns temas que apareceram na busca não eram necessariamente ligados ao interesse do nosso trabalho, utilizada como descritor, ou seja, as IDP.

Com base nisto, partimos para apurar nosso procedimento, através da adoção de limites de busca ***("****and NOT hiv and NOT acquired immunodeficiency syndrome and NOT virus and NOT cancer NOT htlv and NOT AIDS")*, resultando em 362 artigos (**Figura 2**)

Observamos que ainda existiam alguns trabalhos sobre a Síndrome de DiGeorge, uma síndrome de imunodeficiência primária bem reconhecida e cujas manifestações incluem defeitos do sistema imune além de anormalidades em muitos outros órgãos e sistemas e estes relatos não eram necessariamente informativos sobre aspectos de infecção e imunidade. Optamos, então, pela exclusão desta síndrome (*AND NOT DiGeorge, AND NOT 22q*), restando um total de 357 artigos.

Na análise dos 357 artigos, 172 artigos foram excluídos, por não estarem relacionados com nenhuma IDP e 64 por não serem estudos de relatos de casos (detalhados na figura 2), sendo selecionados para analise integral, 124 artigos.

Nesta busca realizada, o que mais nos chamou a atenção foi o fato de que poucos relatos de caso foram recuperados, inclusive das imunodeficiências onde as manifestações orais são sabidamente parte do quadro clínico apresentado pelos pacientes, o que nos levou a questionar se o procedimento de busca era eficiente para recuperar essa categoria de trabalhos.

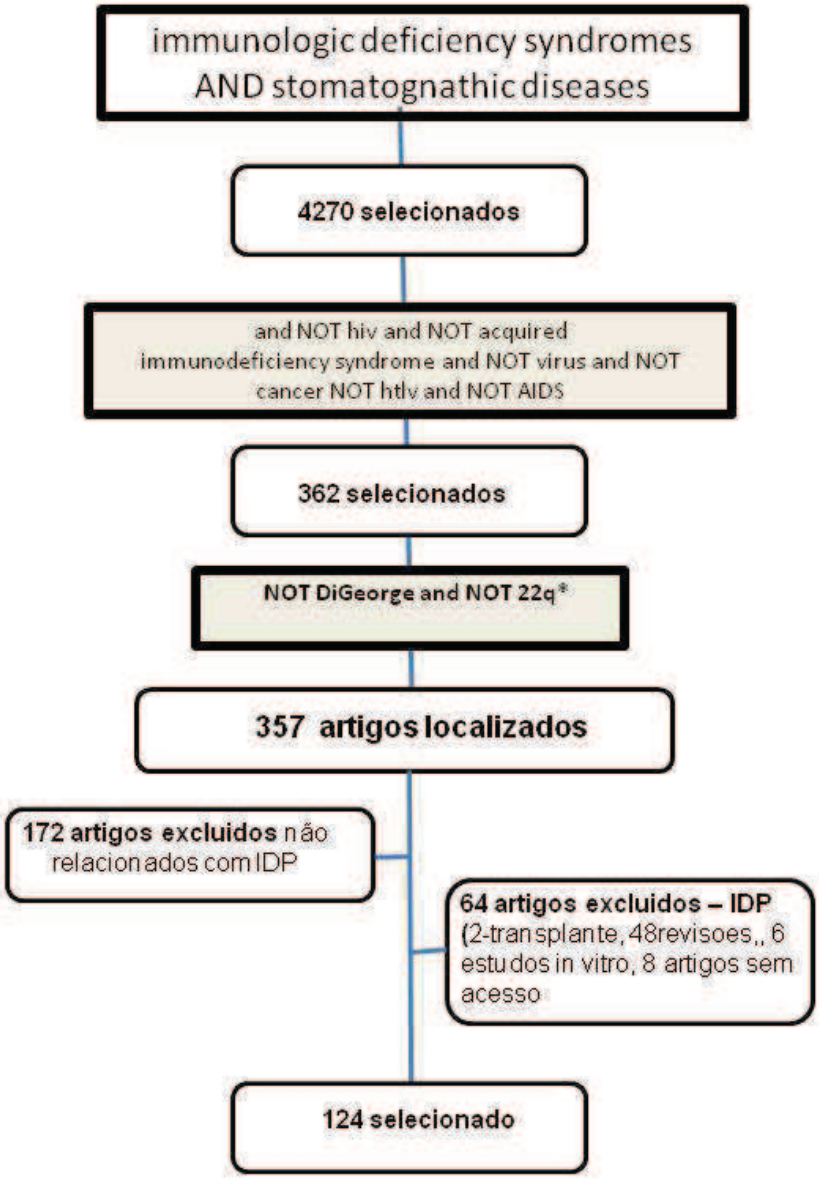
Com o objetivo de evitarmos a perda de referências importantes, modificamos nossa estratégia de busca, partindo para a busca de relatos de casos de IDPs, e, dentro dos artigos recuperados, procuramos sistematicamente a menção de alterações orais.

Como existem muitas IDPs de vários grupos, o trabalho envolvido aumentou muito, e, portanto, tomamos a decisão de limitar nossa busca ao grupo de defeitos de fagócitos, que sabidamente apresentam manifestações orais e para os quais é possível estabelecer relações entre fisiopatologia e clínica.

Tendo, portanto, verificado que a busca por termos abrangentes era inadequada para nossa tarefa, reformulamos nossa estratégia, para assegurar que relatos de casos sobre todos os tipos de IDPs de fagócitos fossem recuperados, fazendo buscas sistemáticas com base nas mutações conhecidas e especificando relatos de casos entre as categorias desejadas de artigos. Os resultados obtidos nas outras buscas são detalhados a partir da seção 6.2, seguindo a ordem elencada na Classificação de IDP (Notarangelo *et al*, 2009) **(Anexo 1).**

Figura 2:

Síndrome de Deficiência Imunológica X Doenças Estomatognáticas



6.2) Neutropenias Congênitas Graves (ELA2 - elastase 2)

A busca realizada na base *pubmed* com os descritores **(*ELA2 and Case Reports*) (Apêndice 1)** localizou um total de 17 artigos. Destes, 9 artigos foram excluídos por não serem relacionados com o gene ELA2 (7 não apresentaram alterações orais e 2 com alterações orais).

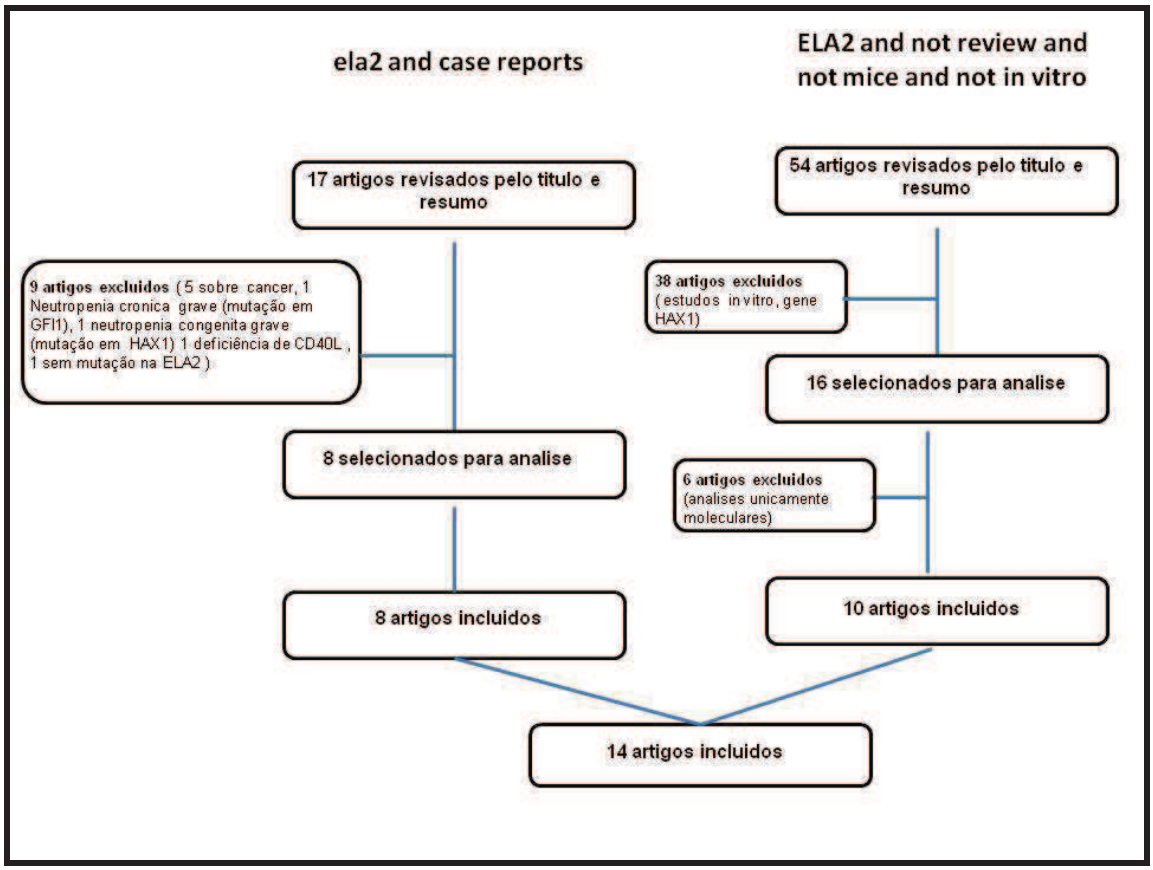
Foram analisados 8 artigos, totalizando 16 pacientes descritos com mutação no gene ELA2 (**Figura 3**). Somente 2 pacientes (12,5%) apresentavam relato de alterações orais; um paciente apresentou inicialmente gengivite e posteriormente periodontite e o outro apresentou úlceras aftosas recorrentes.

Uma segunda busca foi realizada utilizando ***"ELA2 and not review and not mice and not in vitro"*** **(Apêndice 2),** com o objetivo de se obter uma busca mais abrangente. Nessa busca encontramos 54 artigos, 44 foram eliminados (estudos de analise *in vitro*, estudos de análises exclusivamente moleculares e análises de outros genes). Desta segunda busca foram analisados 10 artigos, sendo que 6 destes já haviam sido localizados na primeira busca **(Figura 3).** Dos artigos restantes, foram avaliados 24 pacientes, e somente 1 paciente apresentou alterações orais, descritas como estomatite grave e ulcerações de cavidade oral.

Figura 3

Neutropenias Congênitas Graves

(ELA2 - elastase 2)



6.3) Neutropenias Congênitas Graves (GFI1: repressão da Elastase)

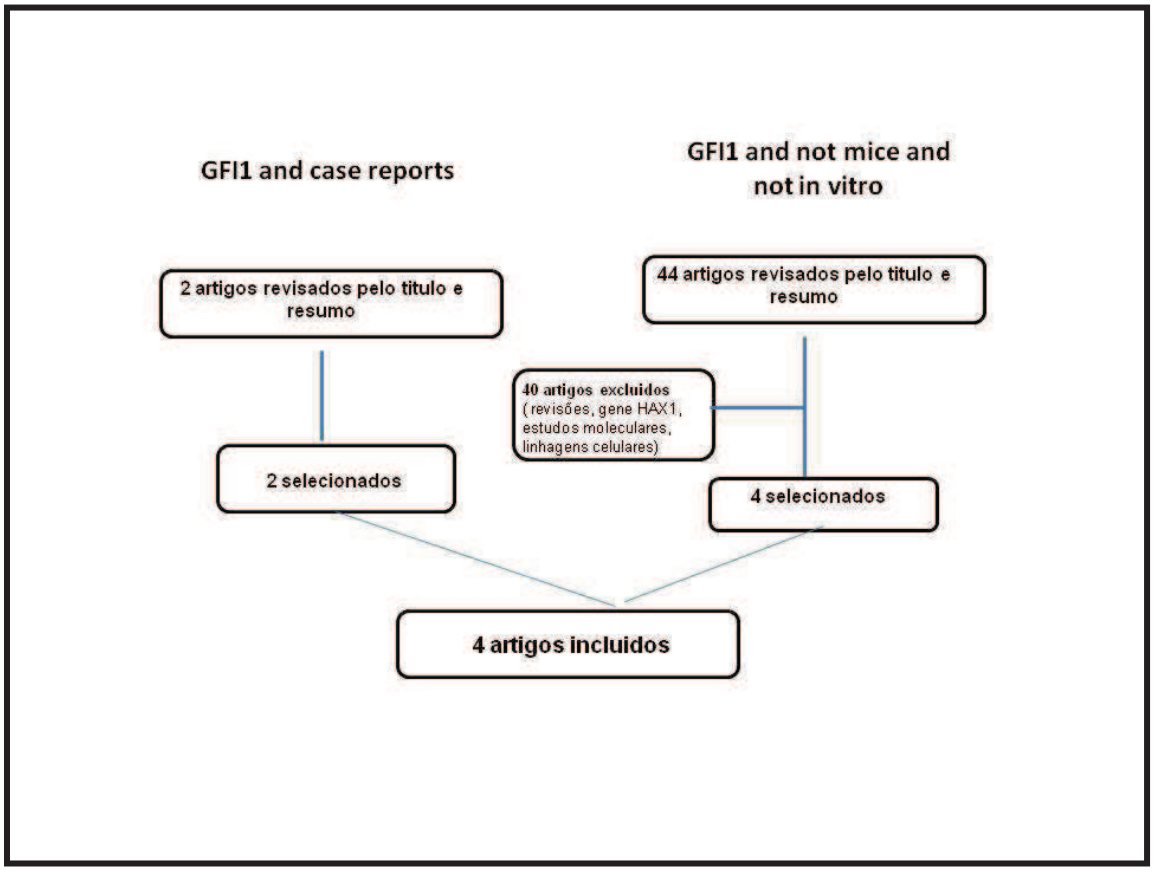
A busca realizada com a descritor *"**GFI1 and case reports**"* identificou 2 artigos **(Apêndice 3)** com descrição de 1 paciente em cada artigo, e em nenhum deles havia relato de alterações orais. Realizamos também uma busca mais abrangente, utilizando somente o descritor **GFI1,** com limite em Humano, excluindo os estudos realizados em camundongo, restando um total de 44 estudos. Porém destes artigos, somente 4 eram análises de casos clínicos, já

encontrados na busca anterior **(Apêndice 4).** Em nenhum deles foi relatada a presença de alterações orais. Dos dois novos artigos não encontrados na primeira busca, um deles era sobre análise molecular do ELA2 e GFI1 em vários pacientes, sem detalhamento da descrição do quadro clínico e o outro era um caso de mutação em ambos os genes ELA2 e GFI1 **(Figura 4).**

Figura 4

Neutropenias Congênitas Graves

(GFI1: repressão da Elastase) (2)



6.4) Síndrome de Kostmann

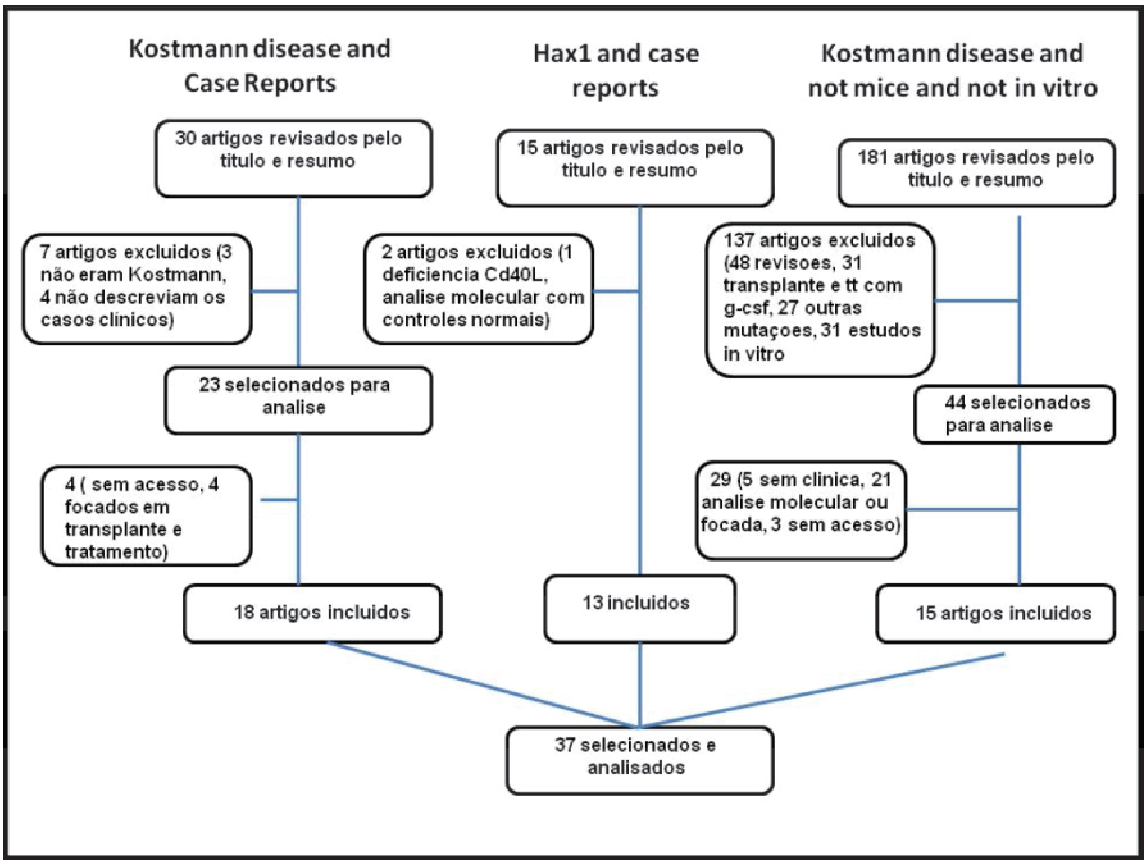
A busca realizada na base *pubmed* com os descritores **(*Kostmann disease and Case Reports*) (Apêndice 5)** localizou um total de 30 artigos. Destes, 8 artigos foram excluídos (3 por não tratarem de Síndrome de Kostmann; 4 artigos que não descreviam os casos clínicos da amostra e 1 artigo por não estar acessível).

Outras duas outras buscas foram realizadas. A primeira busca utilizando os descritores **"*hax1 and case reports*"**, onde foram localizados 15 **(Apêndice 6),** 2 foram excluídos e 13 seguiram para analise. A segunda busca com os descritores ***"Kostmann disease and not mice and not in vitro"*** localizando 181 artigos **(Apêndice 7).** Na primeira busca, todos os artigos foram incluídos para análise. Já na segunda busca, do total de 181 artigos, foram eliminados 137 (48 revisões, 31 artigos sobre transplante e tratamento com G-CSF, 27 sobre outras mutações, 31 estudos in vitro; além da eliminação de 29 artigos sendo 5 que não apresentaram descrição clínica, 21 sem analise molecular e 3 por falta de acesso ao artigo).

Após a remoção das duplicidades encontradas nas três buscas realizadas com os diferentes descritores, a análise foi feita nos 37 artigos de relatos de caso de doença de Kostmann, detalhados na **Figura 5**. No universo amostral de 77 pacientes analisados, 57% destes apresentavam relato de manifestações orais. As manifestações mais freqüentes foram doença periodontal (68%), aftas (61%) e gengivite (27%). Outras manifestações com menores freqüências foram: Estomatite (20,45%), Candidíase (3%), Sangramento gengival (2%), Perda de osso alveolar (6%), Perda precoce de dentes decíduos (4%), Infecções orais de repetição (2%), Mobilidade dentária (4,55%), Abscesso de glândula parótida (2%), Infecções orais por herpes (2%).

Figura 5

Síndrome de Kostmann (3)



6.5) Deficiência de G6PC3 - Neutropenia com malformações cardíacas e urogenitais

A busca realizada utilizando **"G6PC3" (Apêndice 8)** identificou 17 artigos. Desta análise foram excluídos 12 (6 eram artigos de revisão, 4 eram sobre análises moleculares sem descrição dos relatos de casos, 1 caso tinha diagnostico de câncer e 1 era um estudo em modelo murino).

A segunda busca utilizando *"**G6PC3 and case reports**"* localizou 3 artigos **(Apêndice 9)**, sendo eliminado 1 por não apresentar mutação no referido gene.

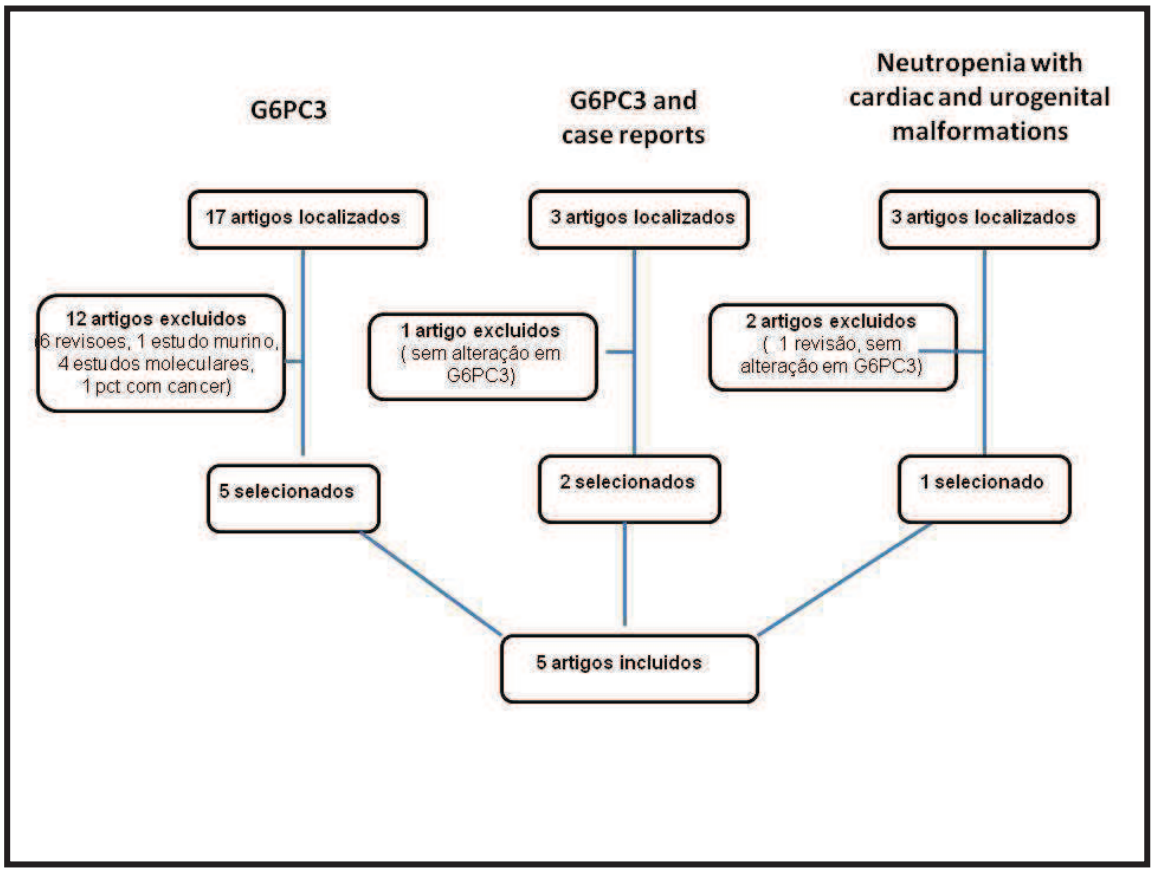
A terceira busca usou os descritores ***"Neutropenia with cardiac and urogenital malformations"*** e encontrou 3 artigos **(Apêndice 10)**, sendo 2 eliminados (1 revisão e 1 por não apresentar mutação em G6PC3).

Foram analisados como resultado destas três buscas, um total de 5 artigos **(Figura 6),** correspondendo a 15 pacientes. Desta amostra, em quatro pacientes houve relato de manifestações orais: Perda precoce de dentes decíduos (n=3), Aftas/úlceras aftosas (n=3), Periodontite (n=2) e Gengivite (n=1).

Figura 6

Deficiência de G6PC3

Neutropenia com malformações cardíacas e urogenitais



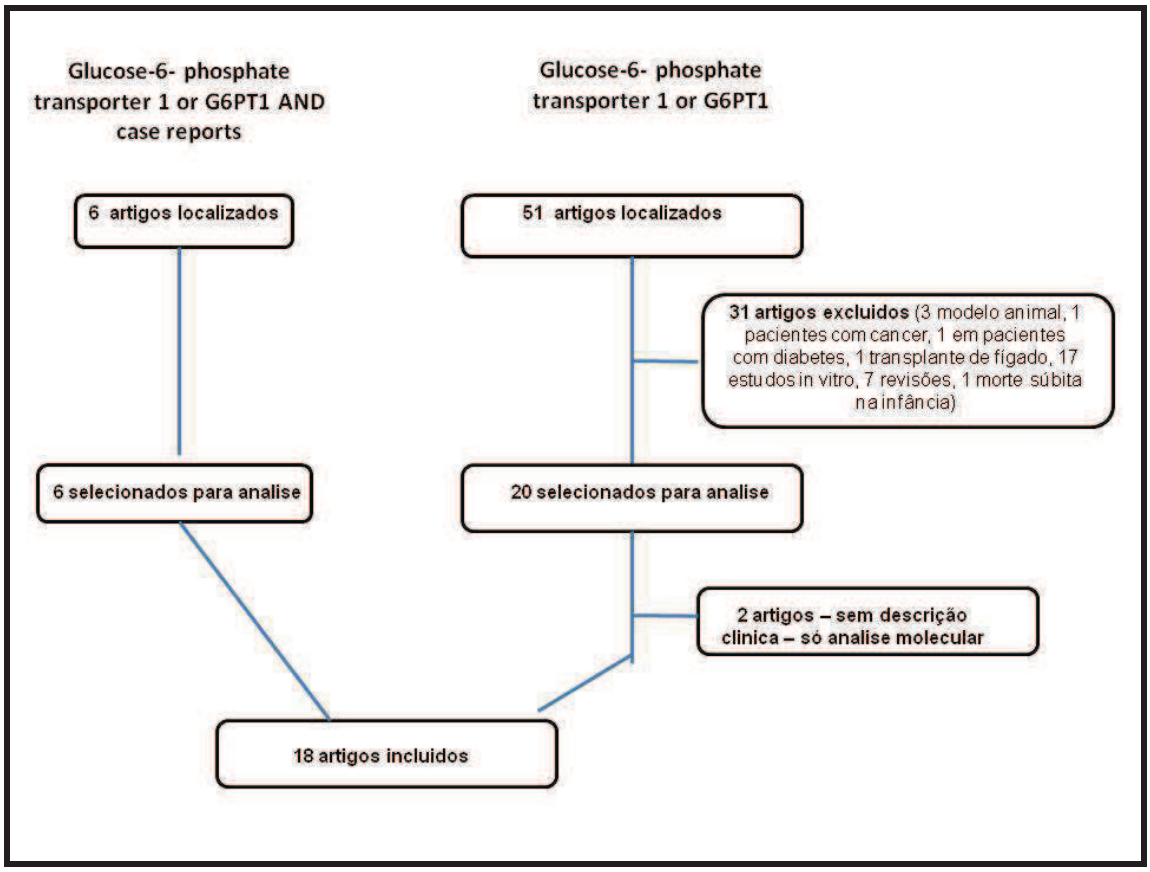
6.6) Doença de estocagem de Glycogenio tipo 1b (gene -G6PT1: Glucose-6-phosphate transporter 1)

A busca realizada utilizando ***"G6PT1 and case reports"*** identificou 6 artigos (Apêndice 11). Optamos por realizar uma busca adicional mais aberta, utilizando o descritor *"**Glucose-6- phosphate transporter 1 or G6PT1**"* com limite em humanos **(Apêndice 12)**. Nessa busca, encontramos 51 artigos, inclusive todos os 6 artigos da busca anterior. Foram excluídos 31 artigos (3 estudos eram em modelo animal, 1 em pacientes com câncer, 1 em pacientes com diabetes, 1 tratava de transplante de fígado, 17 eram estudos *in vitro*, 7 eram artigos de revisão, 1 artigo era em japonês). Um total de 18 artigos foi analisado (**Figura 7**), correspondendo a 143 pacientes com diagnóstico confirmado, e nenhum relato de alteração oral foi encontrado.

Figura 7

Doença de estocagem de Glycogenio tipo 1b

(gene -G6PT1: Glucose-6-phosphate transporter 1)



6.7) Neutropenia Cíclica

A busca realizada utilizando os descritores *"**Cyclic neutropenia syndrome and case reports**"* **(Apêndice 13)** identificou 20 artigos e, destes foram eliminados 19 artigos (14 artigos sobre outras a apresentação de neutropenia em outros tipos de IDP, 2 artigos sobre outras doenças não relacionadas, 1 artigo em russo, 1 estudo *in vitro*, 1 artigo sem acesso).

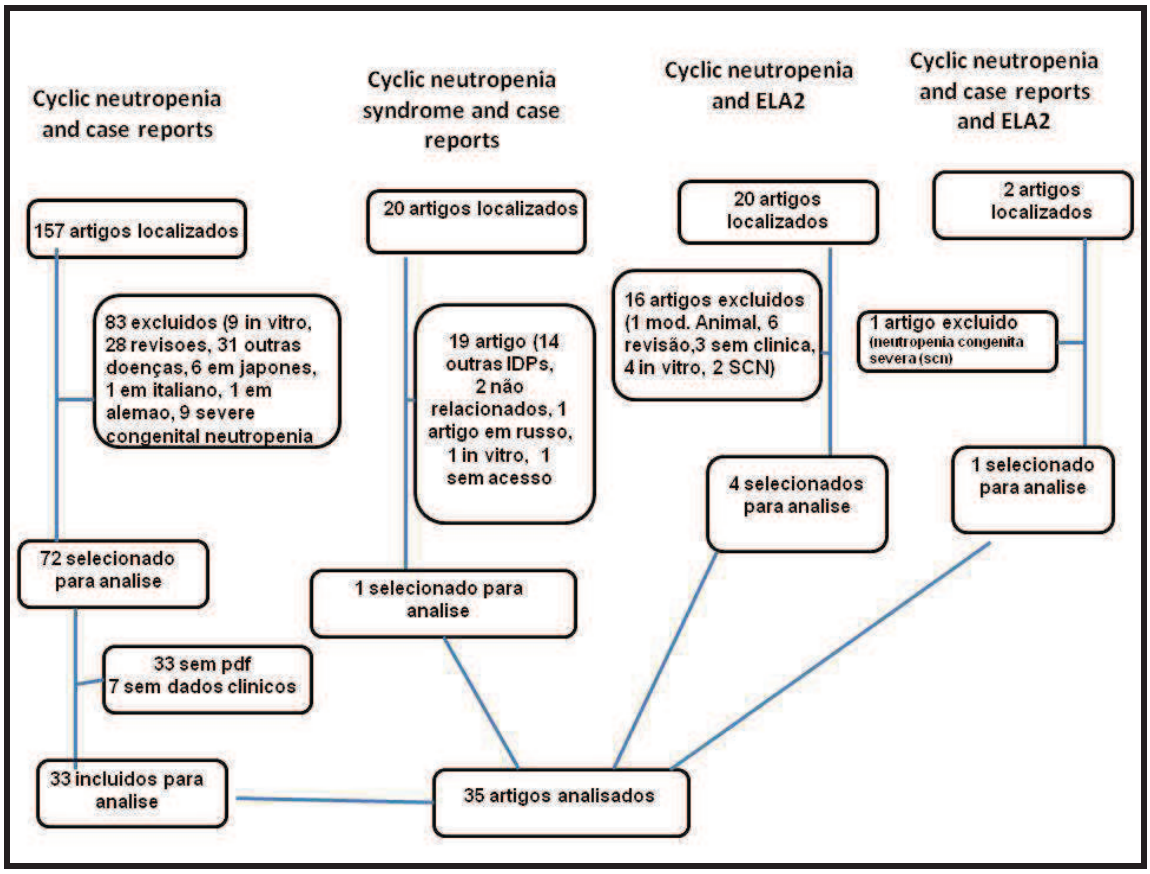
A segunda busca utilizou os descritores *"**Cyclic neutropenia and case reports"*** **(Apêndice 14)** identificando 157 artigos, 83 foram excluídos (28 revisões, 30 artigos sobre outras doenças, 9 estudos *in vitro*, 9 artigos sobre Neutropenia congênita grave, 6 artigos em japonês, 1 em italiano, 1 em alemão). Dentre os 73 artigos selecionados para análise, 38 foram excluídos (32 artigos antigos sem acesso e 7 por não apresentarem descrição dos dados clínicos), resultando em 36 artigos incluídos no estudo.

A terceira e quarta buscas foram baseadas no gene identificados relacionado com a neutropenia cíclica. A terceira busca foi ***"Cyclic neutropenia and ELA2"*** **(Apêndice 15)** que encontrou 21 artigos sendo que 16 artigos foram excluídos (1 em modelo animal, 6 artigos de revisão, 4 artigos sem descrição clinica, 4 estudos *in vitro*, 2 artigos sobre neutropenia congênita grave). A quarta busca utilizou ***"Cyclic neutropenia and case reports and ELA2"*** **(Apêndice 16)** e encontrou 2 artigos, 1 foi excluído por ser sobre neutropenia congênita grave.

Após a eliminação das sobreposições 35 artigos foram incluídos na analise consolidada **(Figura 8)**, totalizando 46 pacientes, 74% (n=34) apresentaram manifestações orais.

Figura 8

Neutropenia Cíclica.



6.8) Neutropenia ligada ao X/ mielodisplasia

A primeiras buscas realizadas utilizaram os descritores ***"X-linked neutropenia AND wasp and case reports"*** **(Apêndice 17)** e **"neutropenia AND wasp and case reports" (Apêndice 18)**, identificaram 2 artigos, sendo que 1 foi eliminado por se tratar de um relato de síndrome de Wiskott-Aldrich.

A segunda busca utilizou os descritores ***"X-linked neutropenia AND wasp"*** **(Apêndice 19)** e identificou 16 artigos, dos quais foram eliminados 10

artigos (2 artigos de revisão, 5 sobre síndrome de Wiskott-Aldrich e 3 estudo *in vitro*).

A terceira busca foi mais abrangente e utilizou os descritores ***"neutropenia and wasp"*** localizando 17 artigos **(Apêndice 20)**. Foram eliminados 12 artigos (9 artigos de revisão, 2 estudo *in vitro*, 1 sobre a síndrome de Wiskott-Aldrich).

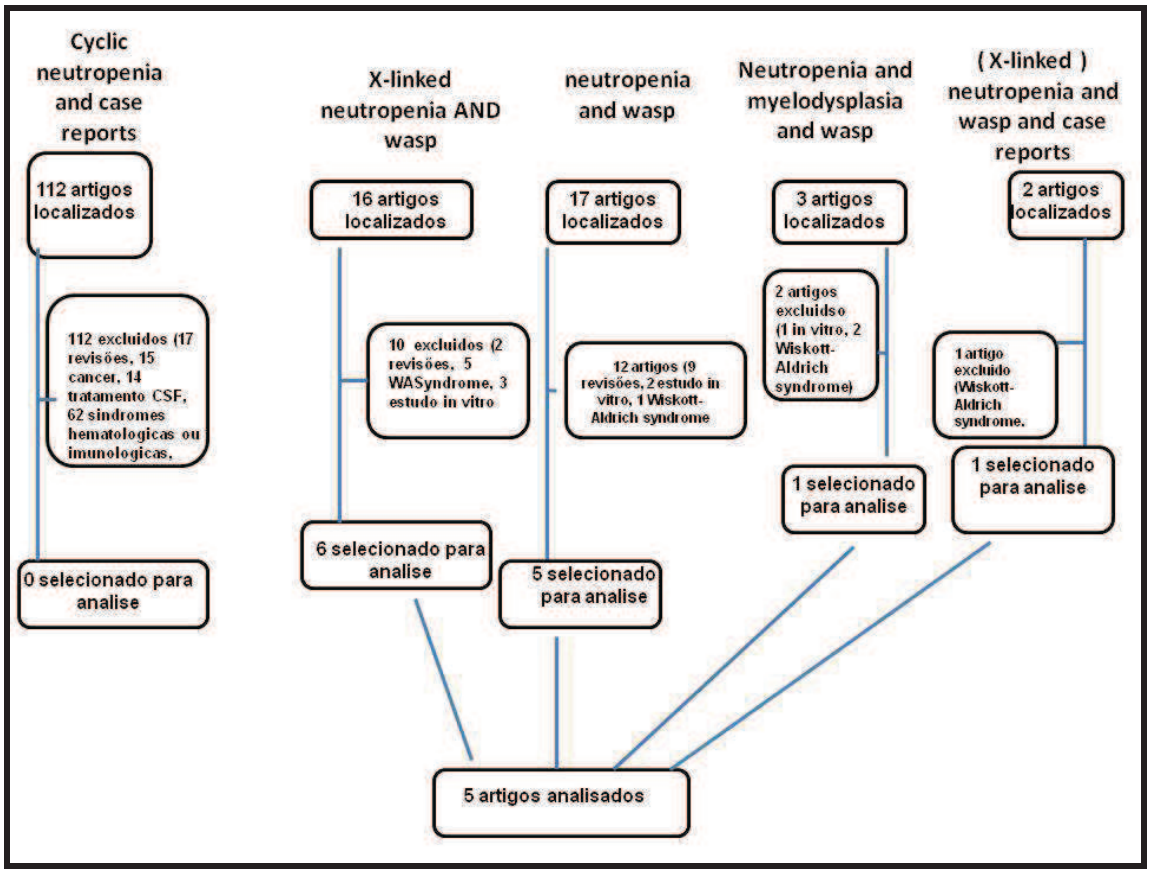
A quarta busca utilizou os descritores ***"Neutropenia and myelodysplasia and wasp"*** localizando 3 artigos **(Apêndice 21)**. Destes, 2 foram eliminados (1 estudo *in vitro*, 2 sobre a síndrome de Wiskott-Aldrich).

A ultima busca foi ***"Neutropenia and myelodysplasia and wasp"*** e localizou 112 artigos **(Apêndice 22)**, dos quais todos foram eliminados (17 artigos de revisão, 15 artigos sobre câncer, 14 sobre tratamento com CSF e 62 de outras síndromes hematológicas ou imunológicas).

Após a eliminação das sobreposições, apenas 5 artigos foram incluídos na análise consolidada **(Figura 9)**, totalizando 41 pacientes. Em nenhum paciente foi descrito relatos de alterações orais. Cabe ressaltar que as descrições clínicas eram bastante resumidas.

Figura 9

Neutropenia ligada ao X/ mielodisplasia



6.9) Deficiência da proteína P14

A busca foi realizada utilizando ***"P14 deficiency syndrome and case reports"*** **(Apêndice 23)**. A outra busca utilizou ***"P14 protein deficiency and case reports"*** **(Apêndice 24)**. Uma outra busca utilizou os descritores ***"Mapbpip protein deficiency and case report"*** **(Apêndice 25)** e a última busca utilizou ***"Endosomal adaptor protein 14 deficiency and case report*** **(Apêndice 26).** Em nenhum destes levantamentos foi identificado qualquer trabalho. Entretanto, ao utilizarmos somente ***"P14 deficiency"*** foram encontrados 17 trabalhos. Destes 17 artigos, 16 artigos foram excluídos (7 artigos por não tratarem da deficiência da proteína 14 e sim de alterações no braço curto do cromossoma (p14), 7 estudos *in vitro* e 2 artigos por se tratarem da posição p14 na coluna vertebral). Restou, portanto, somente 1 artigo sobre a deficiência da proteína 14 **(Figura 10)**, no qual não havia relato de nenhuma alteração oral.

Figura 10

Deficiência da proteína 14

6.10) Deficiência de adesão leucocitária tipo 1

A primeira busca foi realizada utilizando como descritores ***"Leukocyte adhesion deficiency type 1 and case reports"*** **(Apêndice 27)** encontrando 15 artigos e excluído 1 artigo por ser um estudo *in vitro*.

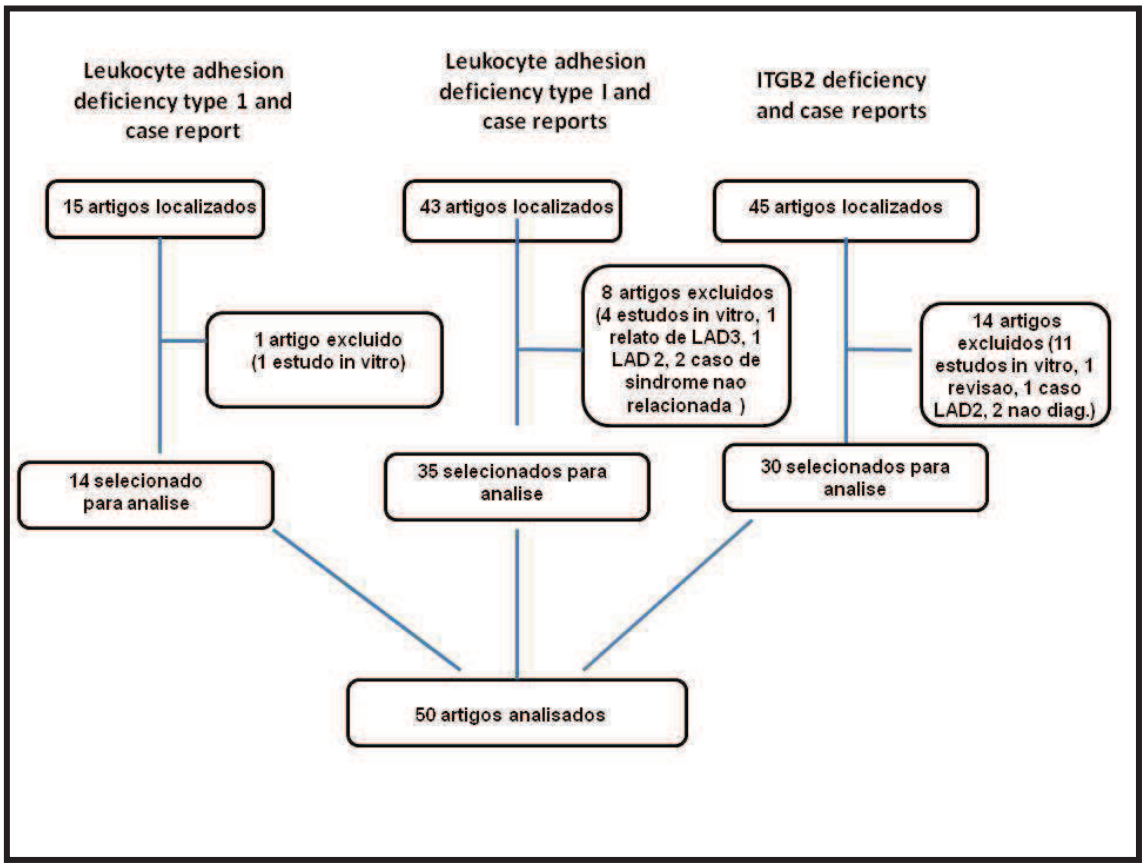
A segunda busca utilizou ***"Leukocyte adhesion deficiency type I and case reports"*** **(Apêndice 28)** encontrando 43 artigos, onde foram excluídos da análise um total de 7 artigos (4 estudos *in vitro*, 1 relato de caso de LAD II, 2 relatos de casos de outras doença).

Na terceira utilizamos o nome do gene envolvido na LAD 1, ***"ITGB2 deficiency and case reports"*** **(Apêndice 29)** localizou 45 artigos, sendo excluídos 14 artigos (11 estudos *in vitro*,1 caso de LAD 2, 2 relatos de caso sem diagnóstico fechado e 1 artigo de revisão).

A análise foi então realizada com 50 artigos após a exclusão das sobreposições **(Figura 11)**, totalizando 80 pacientes. Destes pacientes relatados, 66,25% (N=53) não possuíam relato de nenhuma manifestação oral e 33,75% possuem diversas manifestações orais: 53,3% de doença periodontal, 26% apresentavam gengivite; 13,4% apresentavam mobilidade dentária, perda de osso alveolar e candidíase oral; 6,6% apresentavam úlceras aftosas e sangramento gengival.

Figura 11

Deficiência de adesão leucocitária tipo 1



6.11) Deficiência de adesão leucocitária tipo 2

A primeira busca foi realizada com os descritores ***"Leukocyte adhesion deficiency type 2 and case reports*"** **(Apêndice 30)** encontrando 23 artigos sendo 17 artigos excluídos (15 artigos sobre LAD 1 e 2 sobre outras síndromes).

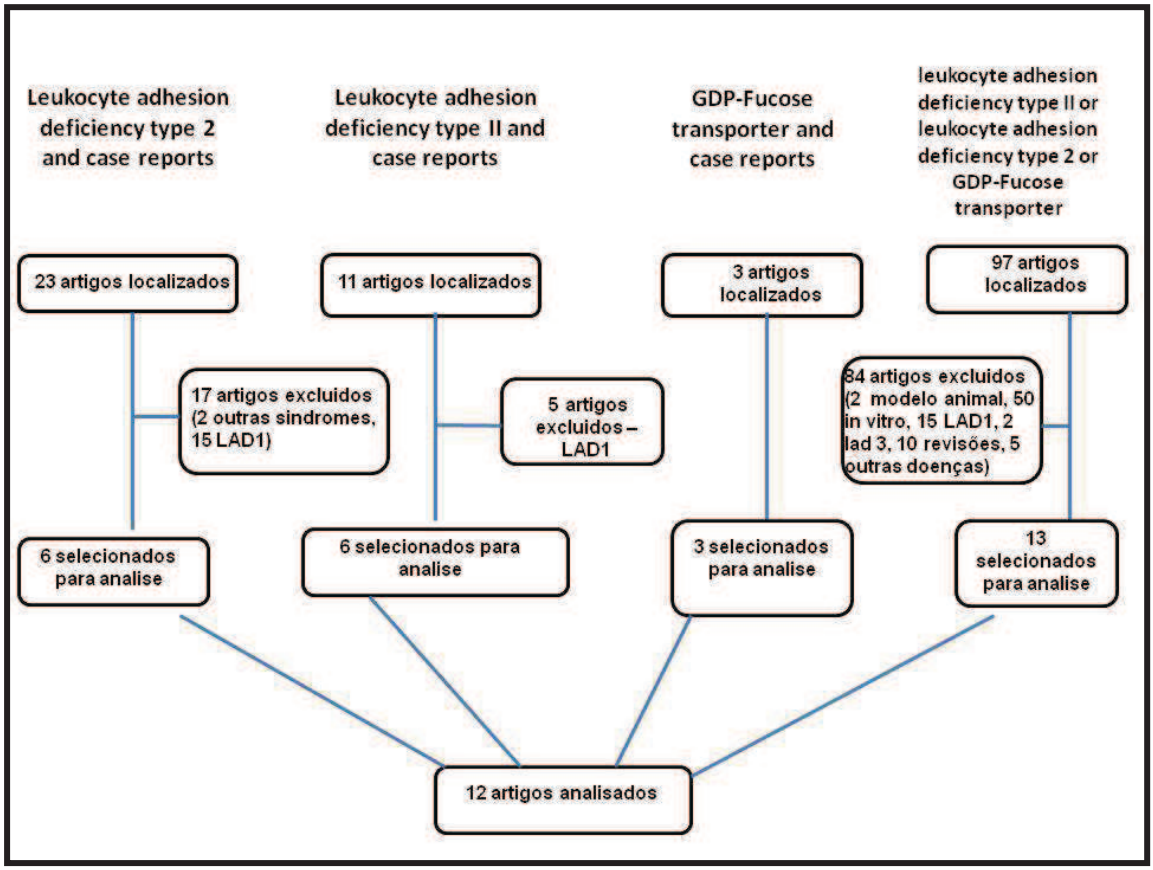
A segunda busca *utilizou* ***"Leukocyte adhesion deficiency type II and case reports"*** **(Apêndice 31)** encontrando 11 artigos e excluídos 5 artigos (5 artigos sobre relatos de caso de LAD I).

Na terceira busca utilizamos o nome do gene envolvido nessa patologia ***"GDP-Fucose transporter"*** **(Apêndice 32)** localizando 3 artigos. Na quarta busca utilizamos ***"Leukocyte adhesion deficiency type II or leukocyte adhesion deficiency type 2 or GDP-Fucose transporter"*** **(Apêndice 33)** localizando 97 artigos. Destes, 84 artigos foram excluídos (2 artigos de modelo animal, 50 estudos *in vitro*, 5 casos de LAD 1, 2 casos de LAD 3, 10 artigos de revisão, 5 artigos sobre outras doenças).

A análise foi realizada, após a exclusão das sobreposições, em 12 artigos **(Figura 12)** e em 21 pacientes. Desta amostra, 52% (N=11) não possuíam nenhuma manifestação oral descrita e 48% possuíam diversas manifestações orais: em 100% dos casos, os pacientes apresentaram doença periodontal e 10% possuíam história de perda precoce de dentes decíduos.

Figura 12

Deficiência de adesão leucocitária tipo 2



6.12) Deficiência de adesão leucocitária tipo 3

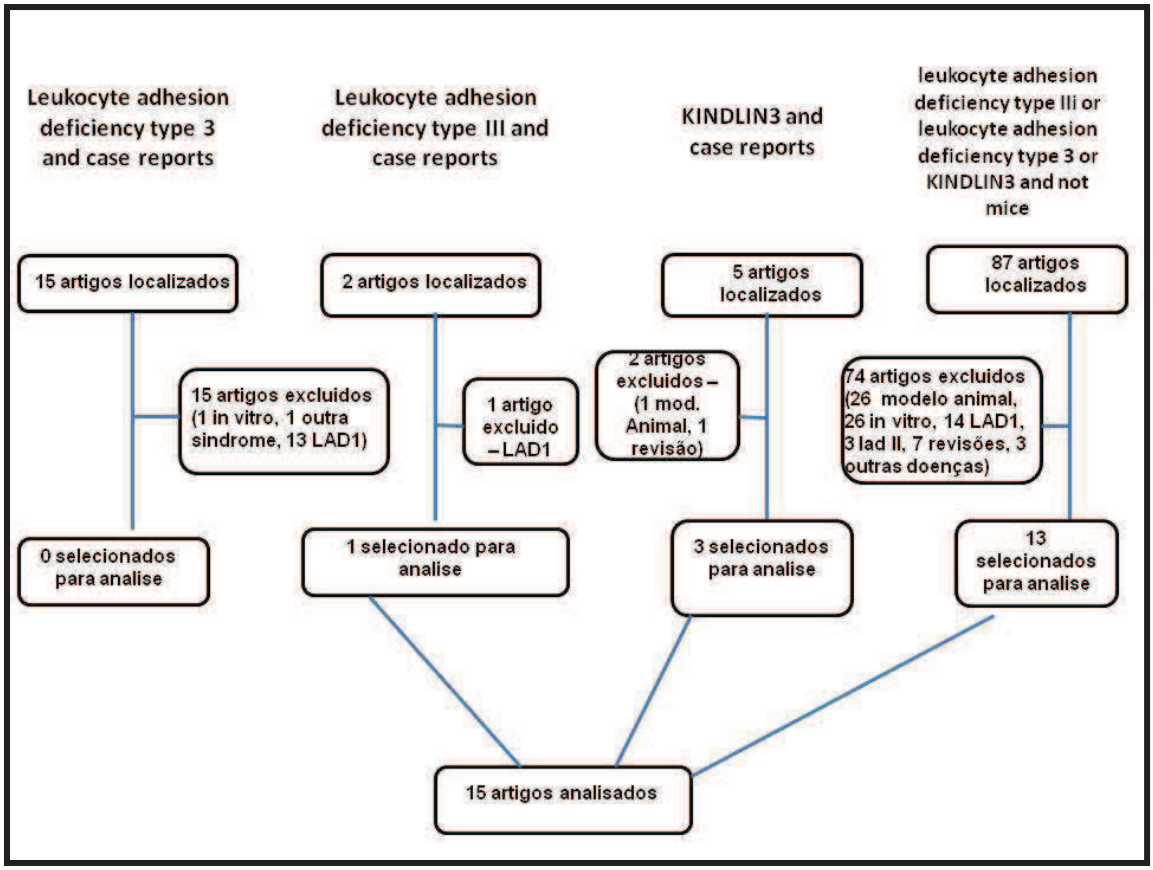
A primeira busca foi realizada com os descritores ***"Leukocyte adhesion deficiency type 3 and case reports"*** **(Apêndice 34)** encontrando 15 artigos e a exclusão de todos estes artigos (1 estudo *in vitro*, 1 artigo sobre uma outra síndrome, 13 artigos sobre LAD-1). A segunda busca utilizou **"Leukocyte adhesion deficiency type III and case reports" (Apêndice 35)** encontrando 2 artigos, excluindo da analise 1 artigo (relato de LAD I). Na terceira busca utilizamos o nome do gene envolvido nessa patologia

"KINDLIN3 and case reports" **(Apêndice 36)**, localizando 5 artigos e excluindo 2 artigos (1 artigo de revisão e 1 artigo sobre modelo canino).

Na quarta e última busca utilizamos ***"leukocyte adhesion deficiency type III or leukocyte adhesion deficiency type 3 or KINDLIN3"*** **(Apêndice 37)** localizando 87 artigos e excluindo da análise 74 artigos (26 modelo animal, 41 in vitro, 14 LAD1, 3 lad II, 17 revisões, 3 outras doenças).

A analise foi realizada nos 15 artigos, totalizando 90 pacientes relatodos, após a exclusão das sobreposições **(Figura 13)**. Em um destes artigos, foram analisados 36 pacientes, porém os relatos de casos clínicos não eram individualizados, embora o artigo descrevesse a presença de doença periodontal e cárie. Para a nossa análise, optamos por desconsiderar esses pacientes, sendo incluídos no nosso estudo um total de 52 pacientes. 46 % (N=24) não relataram nenhuma manifestação oral e 54% possuem diversas manifestações orais: Em um artigo 8 dos 9 pacientes relatados apresentavam comprometimento oral, mas os autores não detalharam as alterações encontradas; Sangramento gengival esteve presente em 14,3%, 10,7% apresentavam candidíase, 7% periodontite.

Figura 13
Deficiência de adesão leucocitária tipo 3



6.13) Deficiência de Rac2

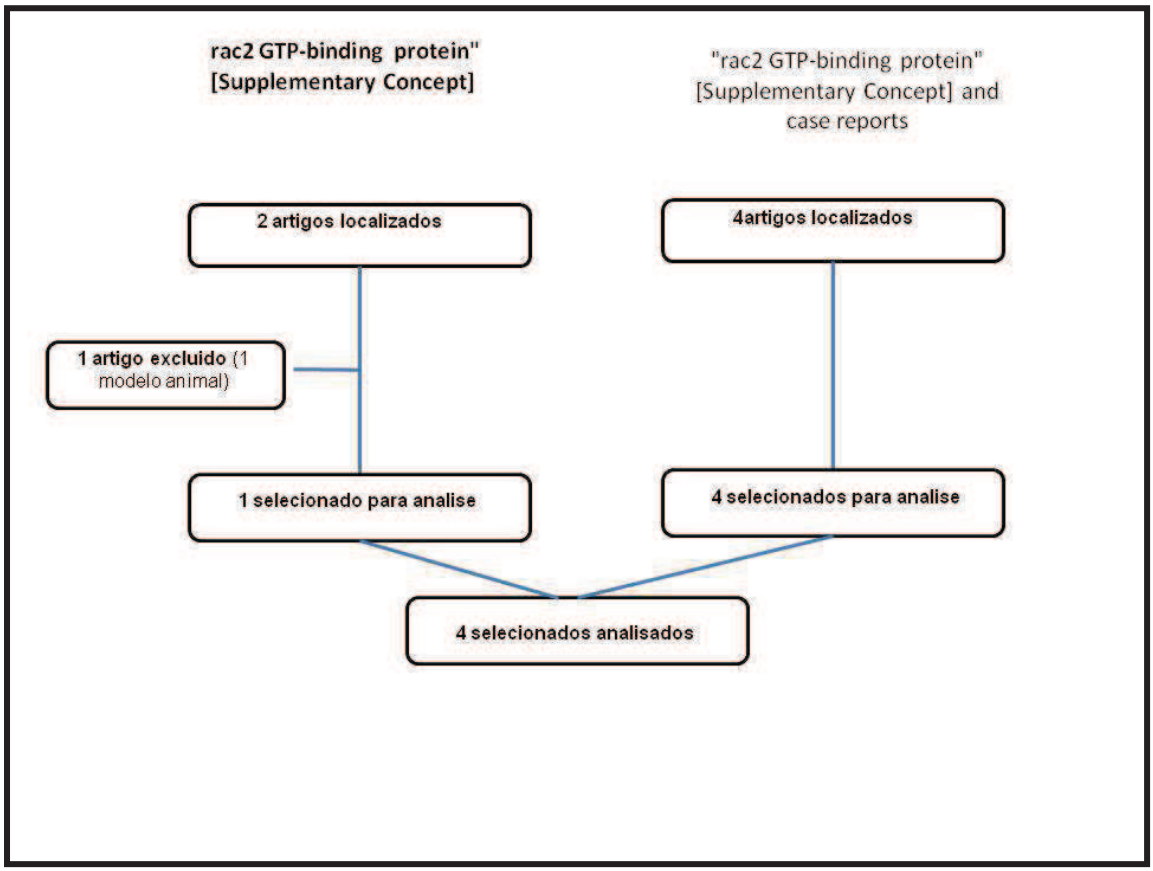
A busca realizada utilizando os descritores *MeSh* ***"rac2 GTP-binding protein [Supplementary Concept] and case reports"*** **(Apêndice 38)** apresentou 4 artigos. Realizamos também uma busca mais aberta utilizando-se a denominação estabelecida na classificação das IDP (Notarangelo *et al*, 2009), ou seja, ***"Rac 2 deficiency and case reports"*** **(Apêndice 39)**,

encontramos 2 artigos (1 artigo que havia sido encontrado na primeira busca e o segundo que foi excluído por não possuir relação com doença).

Os 4 artigos incluídos para análise, totalizaram 4 pacientes e em nenhum destes casos houve relato de alterações orais, possivelmente pela idade muito precoce dos pacientes (inferior ou igual a 1 ano) **(Figura 14).**

Figura 14

Deficiência de Rac 2



6.14) Deficiência de □-Actina

A busca realizada utilizando ***"b-Actin deficiency and case reports"*** **(Apêndice 40)** não localizou nenhum artigo. Fizemos, então, uma segunda busca utilizando ***"Cytoplasmic actin deficiency and case reports"*** **(Apêndice 41)** que localizou 3 artigos, porém nenhum destes estava relacionado com a deficiência de β-Actina. Uma terceira busca foi realizada utilizando ***"ACTB and case reports"*** **(Apêndice 42)** e nenhum artigo foi encontrado.

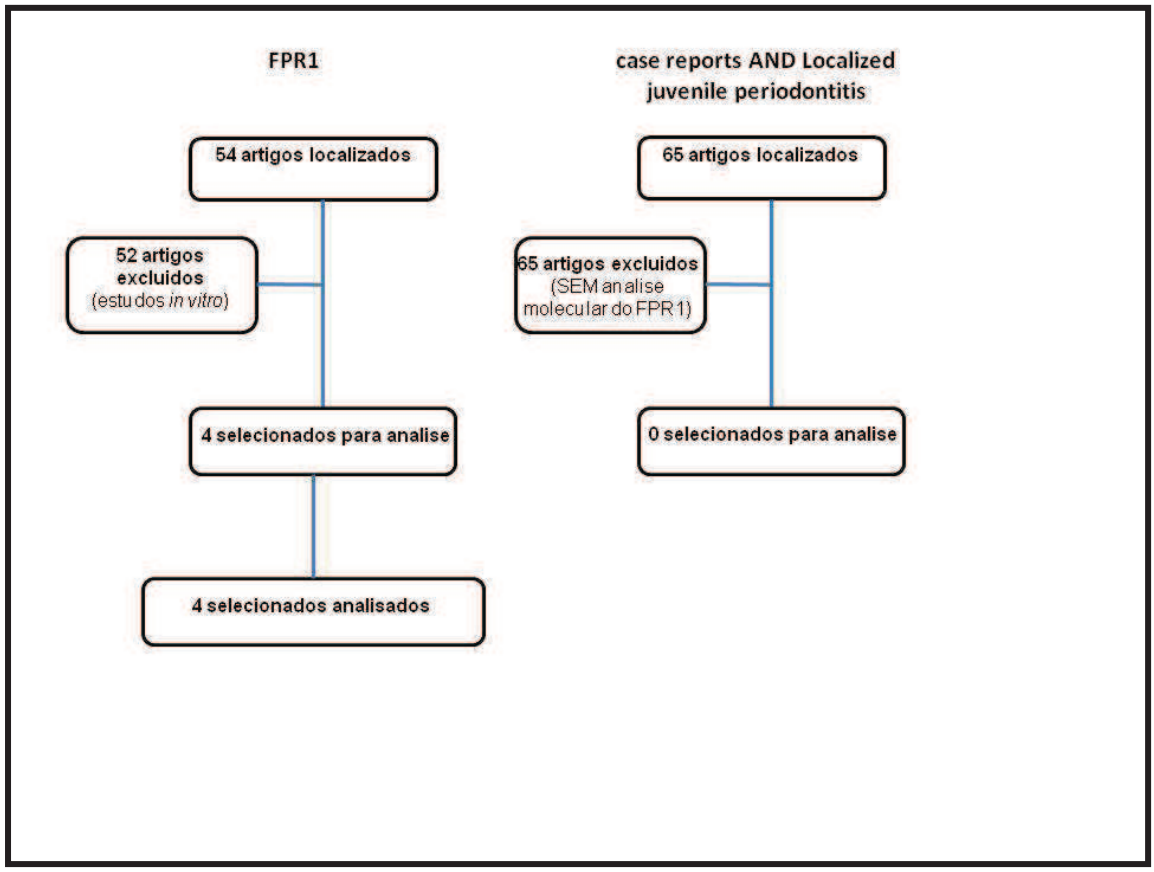
6.15) Periodontite juvenil localizada – Formylpeptide FPR1: Chemokine receptor

A busca realizada com os descritores ***"case reports AND Localized juvenile periodontitis"*** **(Apêndice 43)** encontrou 65 artigos, mas nenhum deles realizava a análise molecular do FPR1. Uma nova busca foi realizada utilizando o descritor ***"FPR1 protein, human"*** **(Apêndice 44)**, encontrando 56 artigos e foram excluídos 52 artigos (52 eram estudos *in vitro* de vários aspectos). 4 estudos de coortes eram com pacientes com periodontite

Foram analisados 4 artigos de estudos de coortes, totalizando 223 pacientes com periodontite localizada ou agressiva aonde a expressão ou o polimorfismo de base única (SNP) de FRR1 foram estudados **(Figura 15).**

Figura 15

Periodontite juvenil localizada – Formylpeptide FPR1



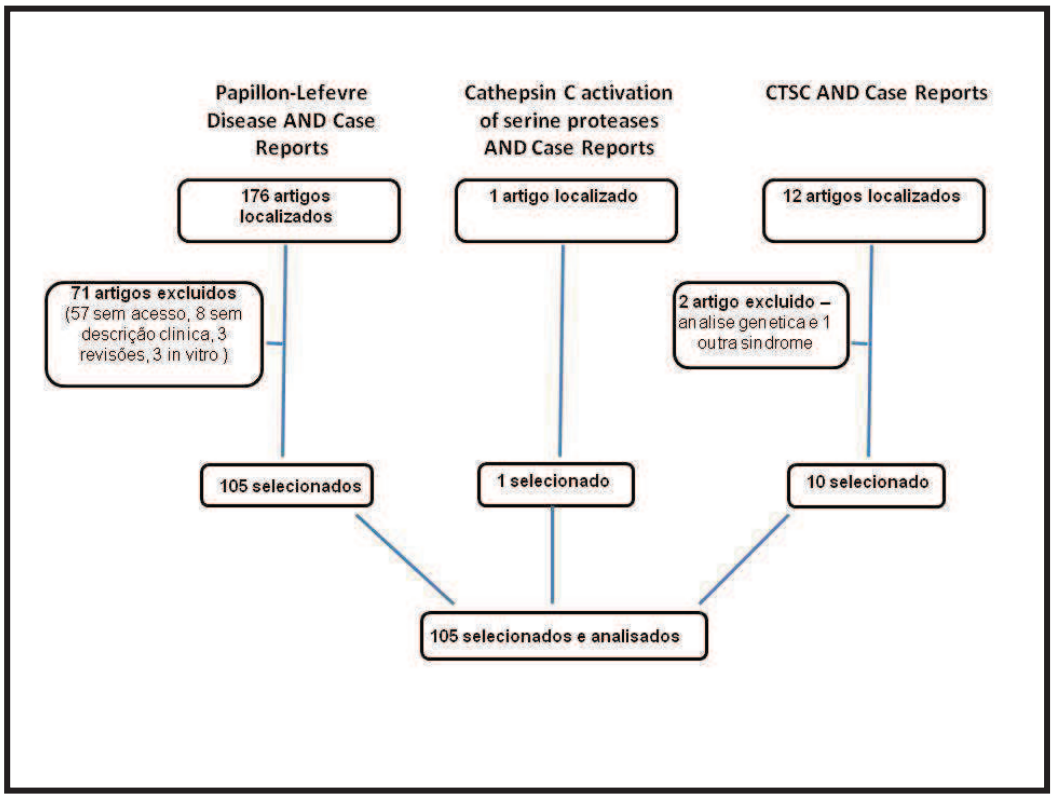
6.16) Síndrome de Papillon-Lefèvre

A busca realizada com os descritores ***'Papillon-Lefevre Disease AND Case Reports"*** **(Apêndice 45)** encontrou 176 artigos e 71 artigos foram excluídos (57 sem acesso aos artigos, 8 sem descrição clínica, 3 revisões, 3 estudos *in vitro)*. A segunda busca realizada utilizou os descritores ***"Cathepsin C activation of serine proteases AND Case Reports"*** **(Apêndice 46)** encontrou 1 artigo e, a terceira busca foi ***"CTSC AND Case Reports"*** **(Apêndice 47)** encontrando 12 artigos, sendo 2 excluídos (1 estudo de análise molecular e outro que não era sobre a Síndrome de Papillon-Lefévre).

A análise foi realizada com 105 artigos depois de excluir as sobreposições, totalizando 152 pacientes **(Figura 16)**. Um total de 8 % não indicaram manifestações orais na descrição clínica, ainda que a doença periodontal faça parte do quadro diagnóstico da síndrome. Em 140 pacientes (92%) houve relatos de manifestações orais: periodontite (53,6%), perda precoce de dente decíduos (42,9%), gengivite (25,0%), perda dentaria (20,7%), perda óssea alveolar (17,9%), mobilidade (17,1%), sangramento gengival (6,4%), abscesso (4,3%), cárie (2,1%), reabsorção óssea mandibular e maxilar (1,4%) e retração gengiva (0,7%).

Figura 16

Síndrome de Papillon-Lefevre



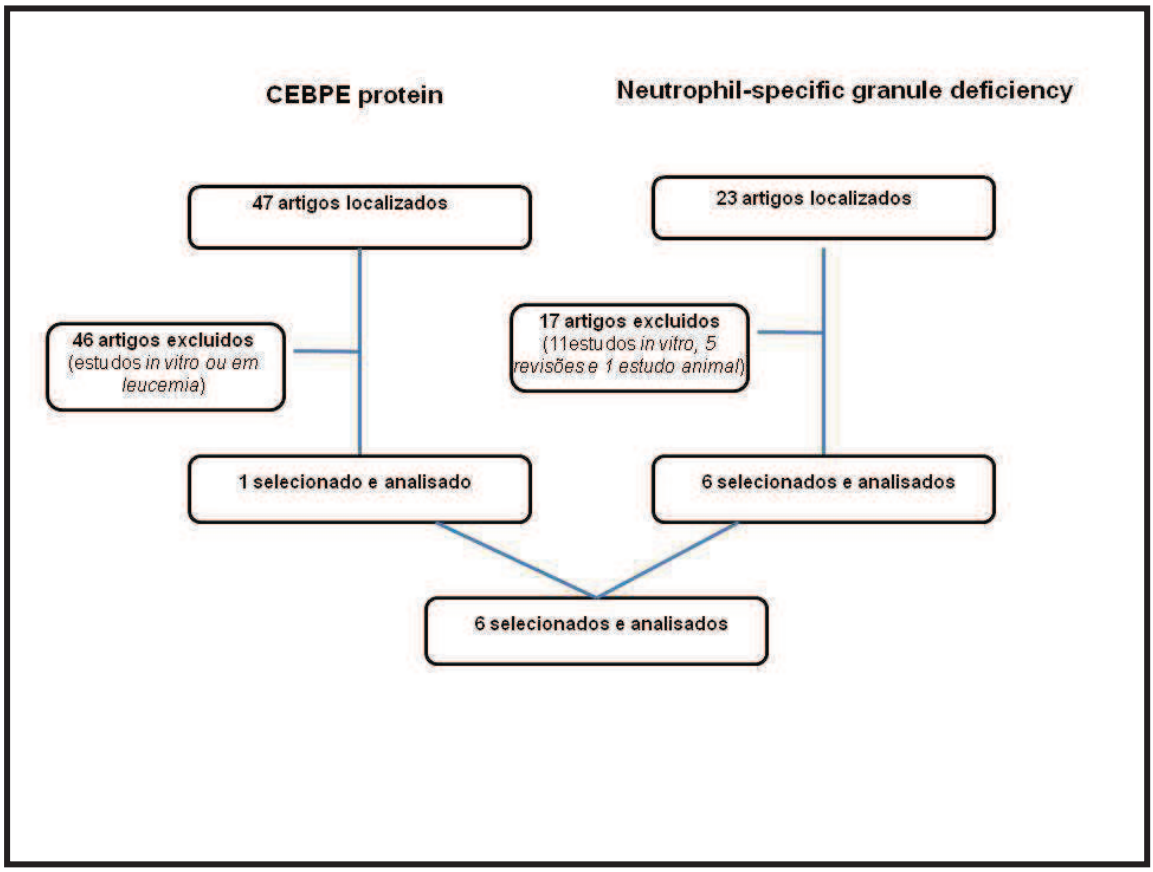
6.17) Deficiência de granulos específicos - CEBPE: (*myeloid transcription factor)*

A busca realizada com o descritor ***"CEBPE protein, human" [Supplementary Concept]''*** **(Apêndice 48)** identificou 47 artigos, dos quais 46 eram estudos com linhagem leucêmicas e caracterização molecular e apenas 1 artigo que descreve um caso de deficiência de granulo especifico, onde foi encontrado um mutação no gene **CEBPε**. A segunda busca foi realizada com o descritor ***"Neutrophil-specific granule deficiency"*** **(Apêndice 49)** onde foram

localizados 23 artigos **(Figura 17)**, 17 foram excluídos por se tratarem de revisão, estudos *in vitro* e modelo animal. No total 6 artigos foram analisados descrevendo 6 pacientes e não houve nenhum relato de manifestação oral.

Figura 17

Deficiência especifica de granulo - CEBPE*: (myeloid transcription factor)*



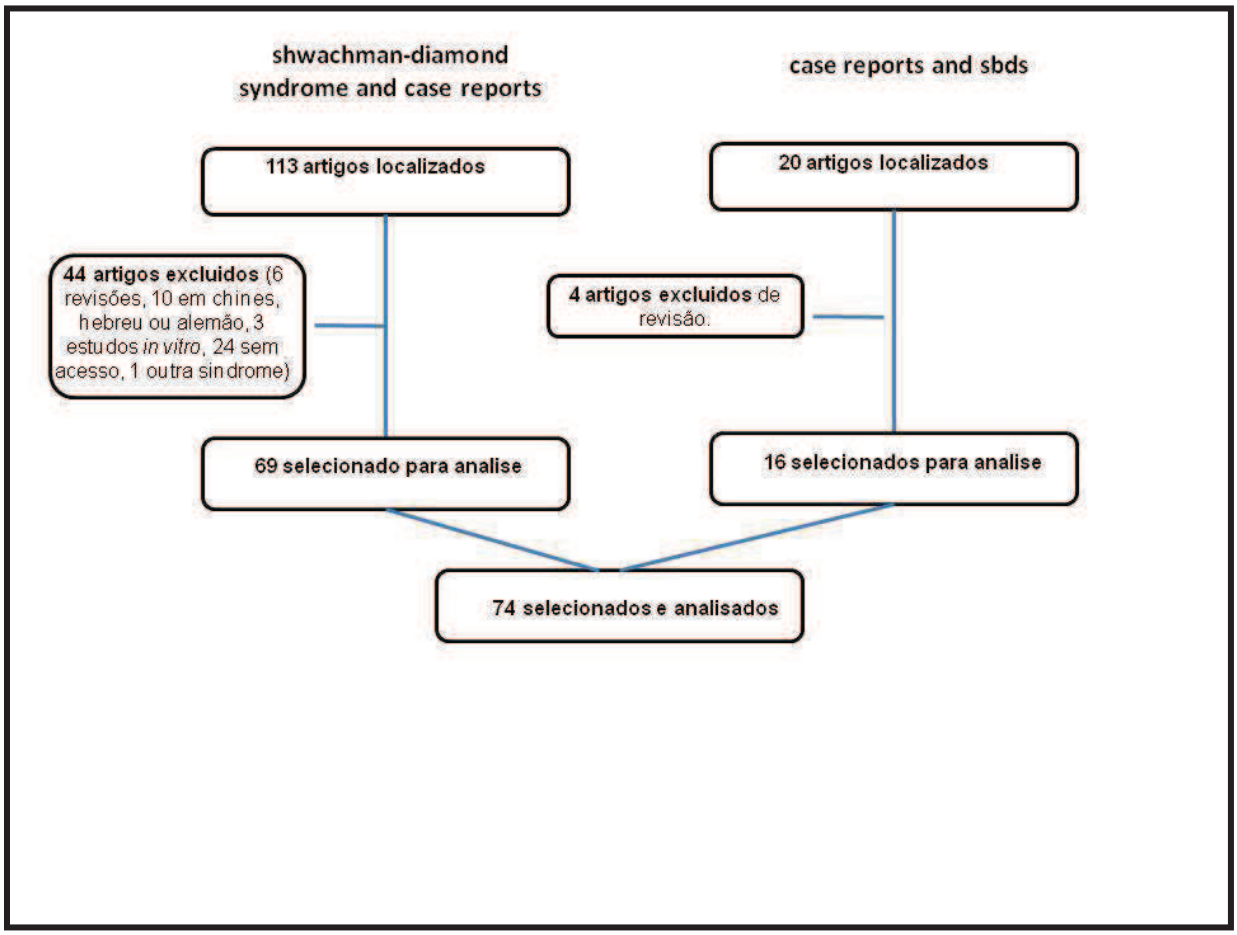
6.18) Síndrome de Shwachman-Diamond.

A busca realizada com o descritor ***"shwachman-diamond syndrome and case reports''*** **(Apêndice 50)** identificou 113 artigos. Foram excluídos 44 (6 revisões, 10 artigos em chines, hebreu ou alemão, 3 estudos in vitro, 24 sem acesso, 1 sendo o relato de outra sindrome). A segunda busca foi realizada com o descritor ***"case reports and SBDS"*** **(Apêndice 51)** onde foram localizados 20 artigos e 4 foram excluídos por se tratarem de revisão.

No total, 74 artigos foram analisados descrevendo 131 pacientes **(Figura 18)**. Não houve relato de manifestação oral para 115 pacientes (87,7%) apesar de manifestações orais estarem presente no quadro diagnóstico dessa síndrome. Apenas 11% apresentaram várias manifestações orais: Anormalidades de desenvolvimento (fenda palatine e fissure labial); cárie em idade precoce; "dentinogênese imperfeita (DI); odontocondrodisplasia; odontodisplasia.

Figura 18

Síndrome de Shwachman-Diamond.



6.19) Doença granulomatosa crônica ligada ao X (CGD)

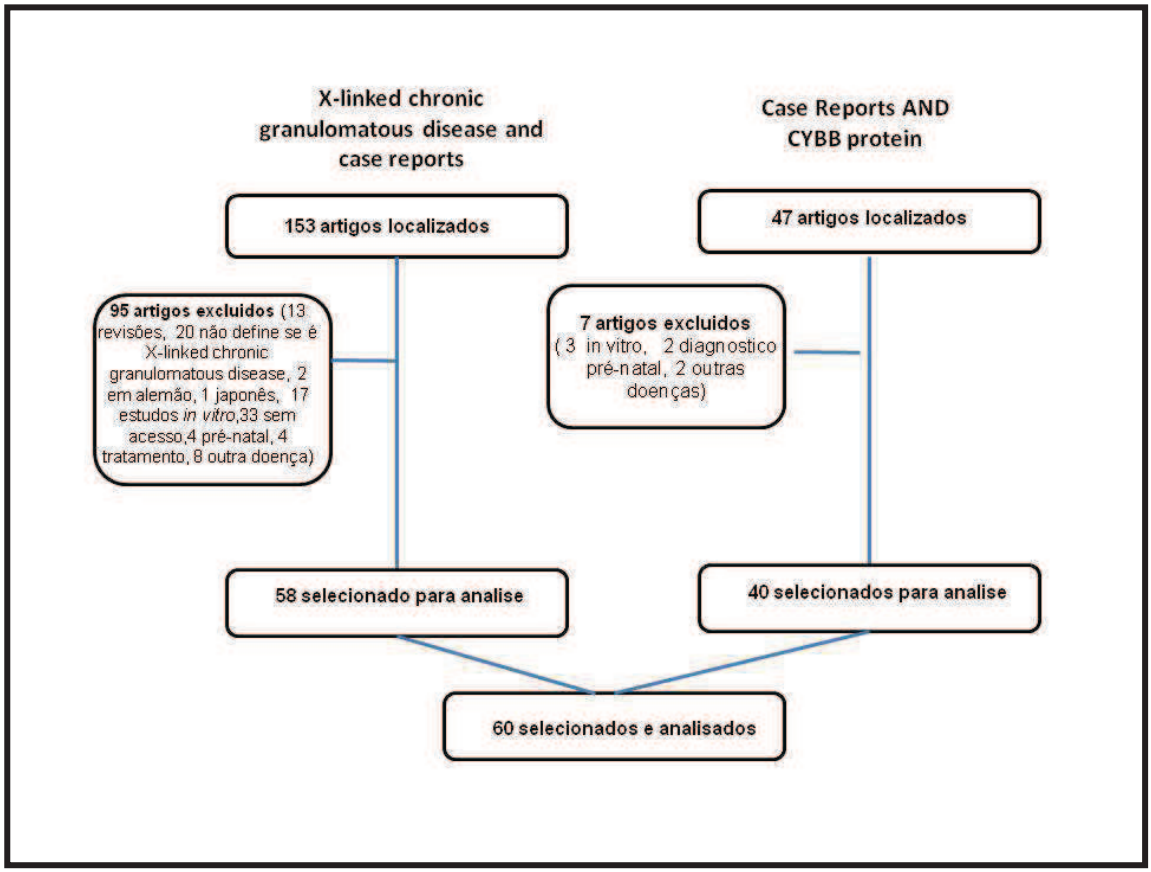
A busca realizada com o descritor ***"X-linked chronic granulomatous disease and case reports''*** **(Apêndice 52)** identificou 153 artigos, foram excluídos 95 artigos (13 revisões, 20 não são Doença granulomatosa crônica ligada ao X, 2 artigos em alemão, 1 em japonês, 17 estudos *in vitro*, 33 sem acesso, 4 estudos em pré-natal, 4 estudos sobre tratamento, 8 sobre outras

doenças). A segunda busca, ***"CYBB protein AND Case Reports"*** **(Apêndice 53)** localizou 47 artigos, 7 artigos foram excluídos.(3 estudos *in vitro,* 2 sobre diagnostico pré-natal e 2 sobre outras doenças). **(Figura 19)**.

A análise foi realizada com 60 artigos depois de excluir as sobreposições totalizando 89 pacientes. Um total de 77 pacientes não apresentou relato de manifestações orais (86,5%). Em 12 pacientes houve relato de manifestações orais: 66,6 % apresentavam estomatite e 33,5 apresenta ulceras orais.

Figura 19

Doença granulomatosa crônica ligada ao X (CGD)



6.20) Doença granulomatosa crônica autossômica (CGD) – CYBA [(Electron transport protein (p22phox)]

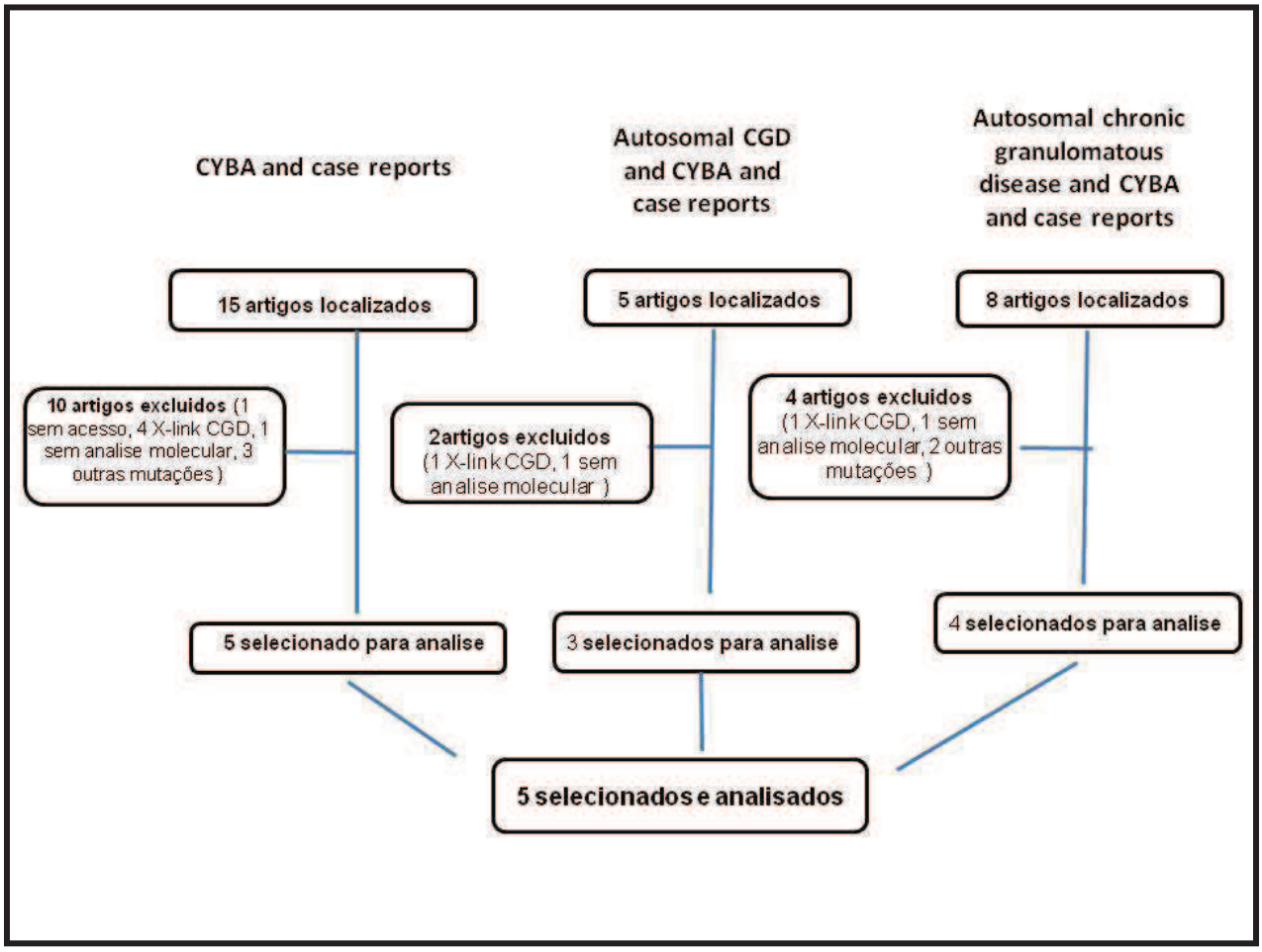
A busca realizada com o descritor "***CYBA and case reports''*** **(Apêndice 54)** identificou 15 artigos, foram excluídos 10 artigos (1 sem acesso, 4 DGC ligado ao X, 1 sem análise molecular, 3 sobre outras mutações). A segunda busca foi baseada nos descritores ***"Autosomal CGD and CYBA and case reports"*** **(Apêndice 55)** selecionado 5 artigos, 2 artigos excluídos (1 de DGC ligado ao X e 1 sem análise molecular). A terceira busca realizada utilizou os descritores ***"Autosomal chronic granulomatous disease and CYBA and case reports"*** **(Apêndice 56)** localizando 8 artigos, sendo que 4 artigos foram excluídos (1 de DGC ligada ao X, 1 sem análise molecular e 2 sobre outras mutações).

A análise foi realizada com 5 artigos depois de excluir as sobreposições totalizando 11 pacientes e nenhuma alteração oral foi relatada. **(Figura 20)**

Em adicional fizemos uma busca utilizando ***"Autosomal chronic granulomatous disease and p22phox and case reports"*** **(Apêndice 57)** foi localizado 1 artigo, já encontrado nas outras buscas, e este foi eliminado por se relativo a Doença granulomatosa crônica ligada ao X, e quando utilizamos os descritores ***"Autosomal chronic granulomatous disease and Electron transport protein and case reports"*** **(Apêndice 58)** localizamos também 1 artigo, que foi excluído por ser uma mutação no Componente p67-phox, não descrita na classificação de IDP utilizada como referência (Notarangelo, 2009) **(anexo 1)**. Foram analisados, portanto, 6 pacientes e não houve relato de alteração oral.

Figura 20

Doença granulomatosa crônica autossômica (CGD) – CYBA



6.21) Doença granulomatosa crônica autossômica (CGD) – NCF1: Adapter protein (p47phox)

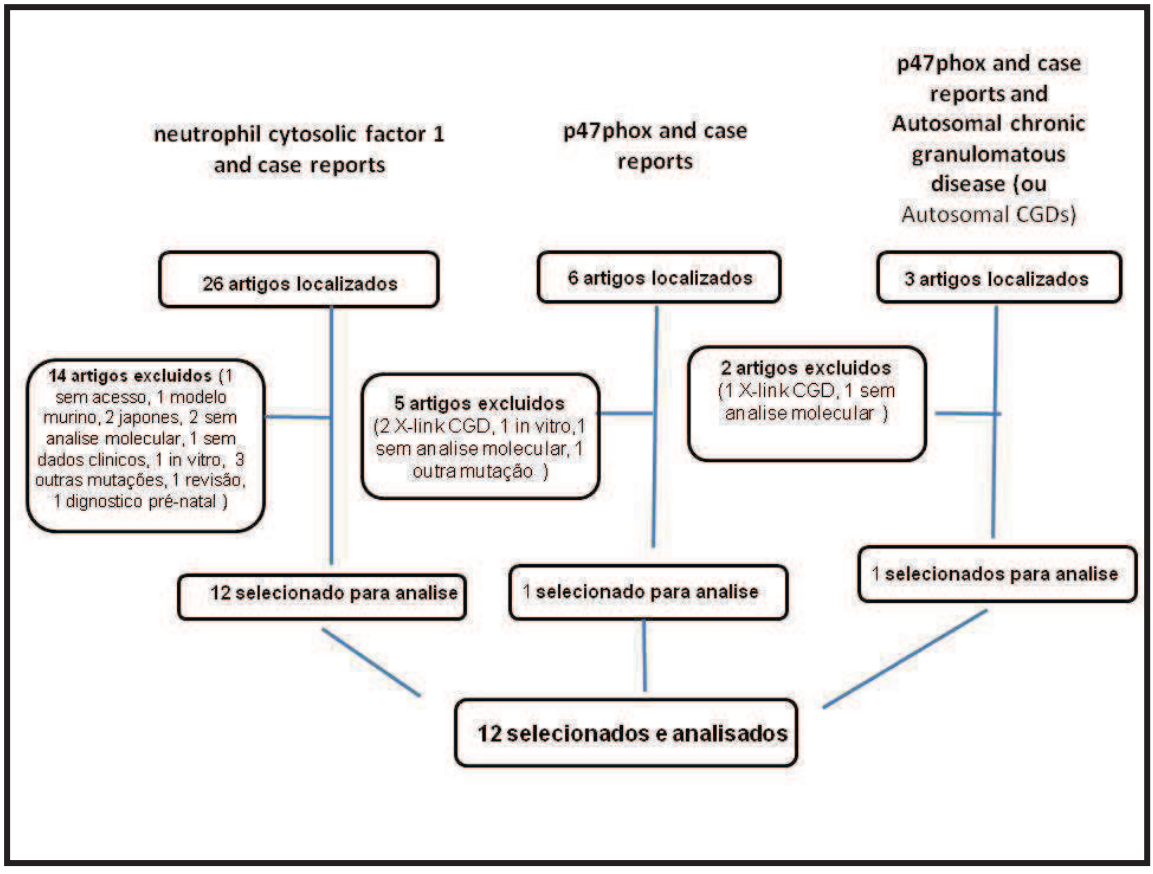
A busca realizada com o descritor **"neutrophil cytosolic factor 1 and case reports" (Apêndice 59)** identificou 26 artigos, foram excluídos 14 artigos (1 sem acesso, 1 modelo murino, 2 em japones, 3 sem analise molecular, 1 sem dados clínicos, 1estudo in vitro, 3 sobre outras mutações, 1 revisão, 1

diagnóstico pré-natal). A segunda busca foi baseada nos descritores **"p47phox and case reports" (Apêndice 60)** localizou 6 artigos e 5 artigos foram excluídos (2 de DGC ligada ao X, 1 estudo *in vitro,*1 sem análise molecular, 1 sobre outra mutação), a terceira busca foi ***"p47phox and case reports and Autosomal chronic granulomatous disease (ou Autosomal CGDs)"*** **(Apêndice 61)** localizando 3 artigos, 2 artigos foram excluídos (1 DGC ligada ao X, 1 sem análise molecular).

A análise foi realizada com 12 artigos depois de excluir as sobreposições totalizando 46 pacientes **(Figura 21**); 91,3% (N=4) não relataram nenhuma manifestação oral, 1 paciente apresentou estomatite e 1 outro apresentou úlceras orais.

Figura 21

Doença granulomatosa crônica autossômica (CGD) – NCF1



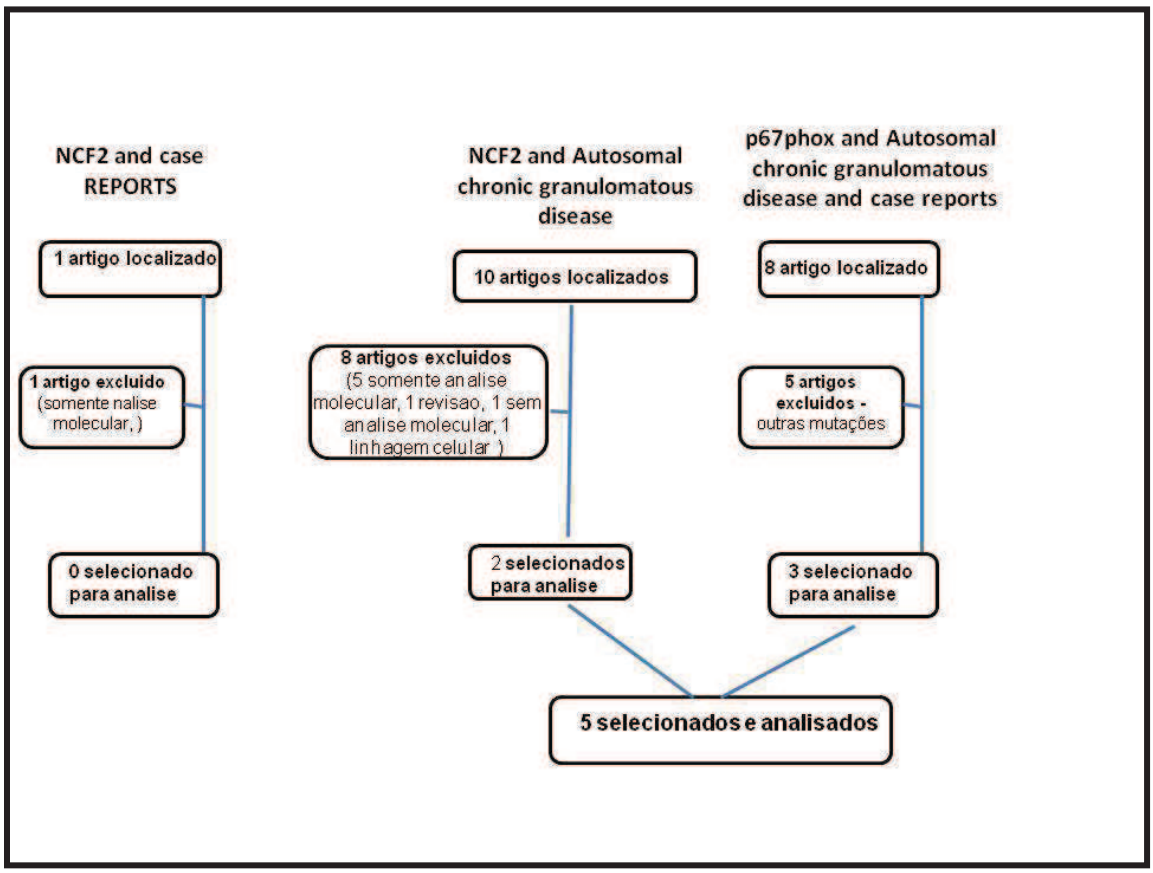
6.22) Doença granulomatosa crônica autossômica (CGD) – NCF2: Activating protein (p67phox)

A busca realizada com o descritor ***"NCF2 and case reports''*** **(Apêndice 62)** identificou 1 artigo que foi eliminado por apresentar somente a análise molecular sem descrever clinicamente o paciente. A segunda busca baseada nos descritores ***"NCF2 and case controls AND Autosomal chronic granulomatous disease"*** **(Apêndice 63)** não encontrou nenhum artigo; a terceira busca foi ***"NCF2 and Autosomal chronic granulomatous disease"*** e localizou 10 artigos **(Apêndice 64)**, sendo que 8 artigos foram excluídos (5 somente realizaram análise molecular, 1 revisão, 1 sem análise molecular, 1 linhagem celular). A última busca foi baseada nos descritores ***"p67phox and Autosomal chronic granulomatous disease and case reports"*** **(Apêndice 65)**, localizando 8 artigos, 5 artigos foram excluídos por analisarem outras mutações.

A analise foi realizada com 5 artigos depois de excluir as sobreposições totalizando 17 pacientes **(Figura 22**) e apenas houve relato de manifestação oral em 1 pacientes com candidíase oral, enquanto que em 94,1% (N=16) não houve relato de nenhuma manifestação oral.

Figura 22

Doença granulomatosa crônica autossômica (CGD) – NCF2



6.23) Deficiência da cadeia β1 do receptor de IL-12 and IL-23

Foram feitas 3 buscas para identificar os relatos de casos de deficiência da cadeia β1 do receptor de IL12 e IL23. A **Figura 23** demonstra a sistemática da busca.

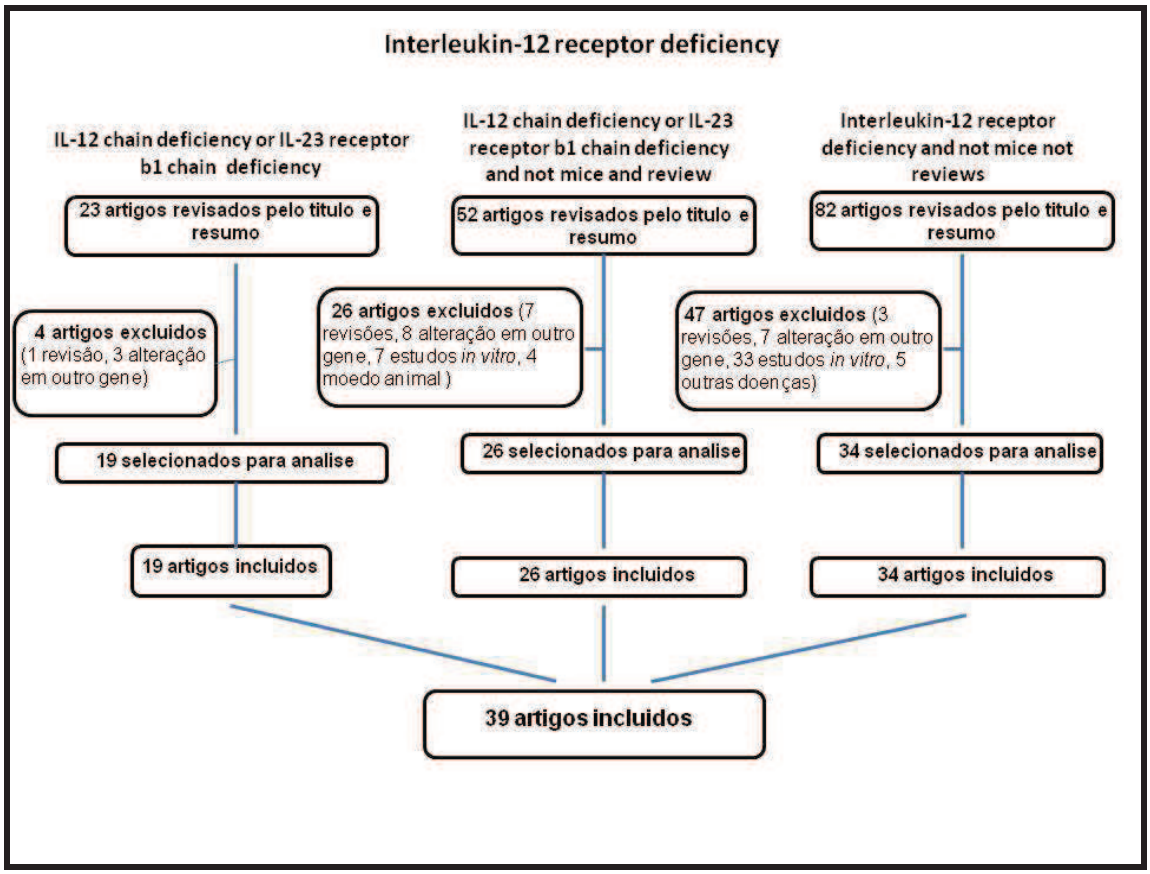
A primeira busca 1 utilizou os descritores ***"IL-12 chain deficiency or IL-23 receptor b1 chain deficiency"*** **(Apêndice 66)** e localizou 23 artigos, sendo 4 artigos excluídos (1 de revisão, 3 sendo sobre alteração em outro gene). A segunda busca utilizou os descritores ***"IL-12 chain deficiency or IL-***

23 receptor b1 chain deficiency and not mice and review" **(Apêndice 67)** e localizou 52 artigos, sendo que 26 artigos foram excluídos (7 artigos de revisões, 8 por se tratarem de alteração em outro gene, 7 estudos *in vitro*, 4 modelo animal). A terceira busca foi ***"Interleukin-12 receptor deficiency and not mice not reviews"*** **(Apêndice 68)** e localizou 82 artigos, sendo 47 artigos foram excluídos (3 revisões, 7 alteração em outro gene, 33 estudos *in vitro*, 5 outras doenças)

Após a exclusão da sobreposição de artigos repetidos nas três buscas foram analisados 39 artigos que totalizavam 245 pacientes. Em apenas um artigo havia uma análise de 141 pacientes de 30 países diferentes, onde a descrição clínica dos casos não estava claramente detalhada. Optamos por excluir este estudo de 141 pacientes. Do restante de pacientes analisados, encontramos 46% sem alterações orais, 45% relatos clínicos insuficientes e 7% apresentaram alterações orais (candidíase oral, infecção por herpes, estomatite, mastoidite causando paralisia do nervo facial).

Figura 23

Deficiência da cadeia b1 do receptor de IL-12 and IL-23



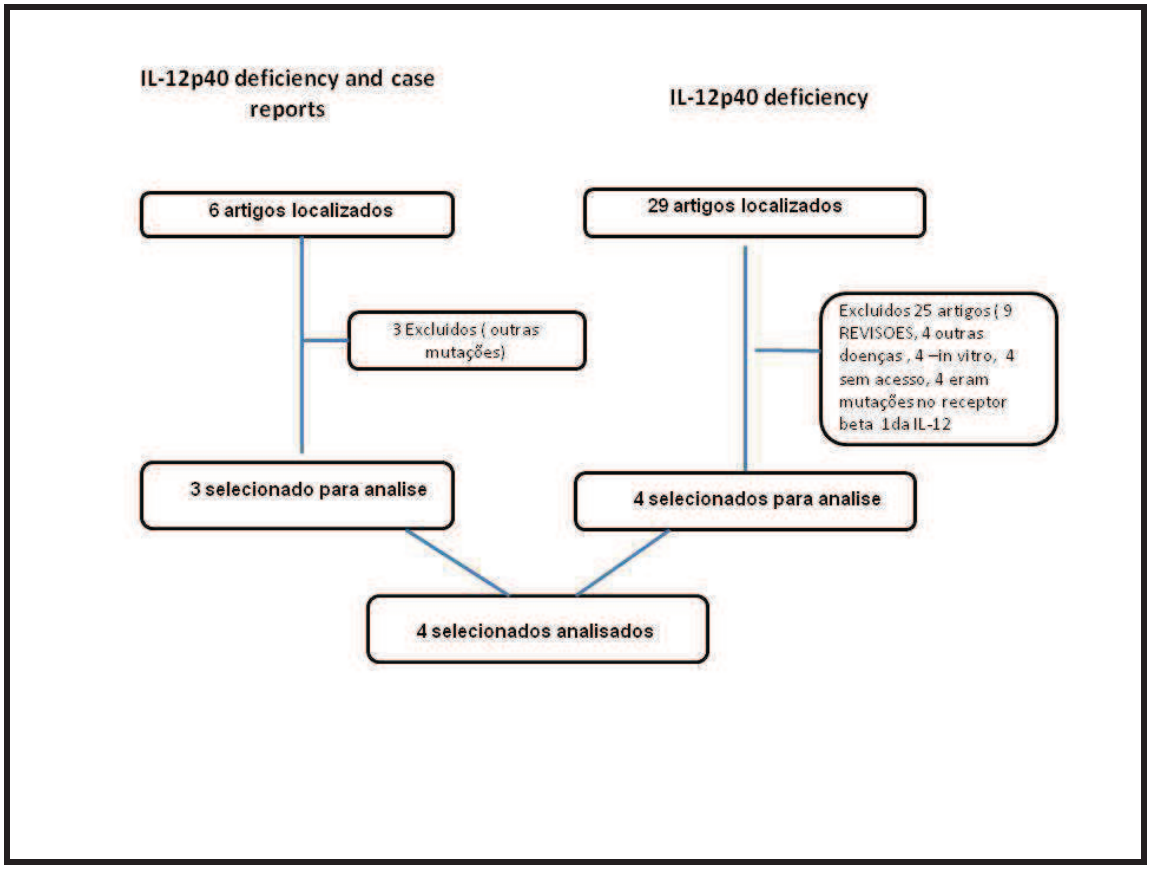
6.24) Deficiência de IL-12p40

A busca realizada com o descritor ***"IL-12p40 deficiency and case reports"*** **(Apêndice 69)** identificou 6 artigos. Foram excluídos 3 artigos por se tratarem de pacientes com outras mutações (STAT5b e IFN-γR1). Nos 3 artigos selecionados foram analisados 3 pacientes e não houve relato de alteração oral.

Uma segunda busca, mais aberta, foi realizada, aonde utilizamos o descritor ***"IL-12p40 deficiency"*** **(Apêndice 70)** e restringindo para estudos murinos. Nessa busca foram localizados 29 artigos, sendo que 25 artigos foram excluídos (4 revisões, 4 relatos de pacientes com outras doenças não relacionadas como tuberculose e lupus, por exemplo, 3 artigos de pacientes portadores de mutações nos genes STAT5b, IFN-γR2 e IFN-γR1, 4 estudos *in vitro*). **(Figura 24).** Um total de 4 artigos foram analisados totalizando 18 pacientes e não houve nenhum relato de manifestações orais.

Figura 24

Deficiência de IL-12p40



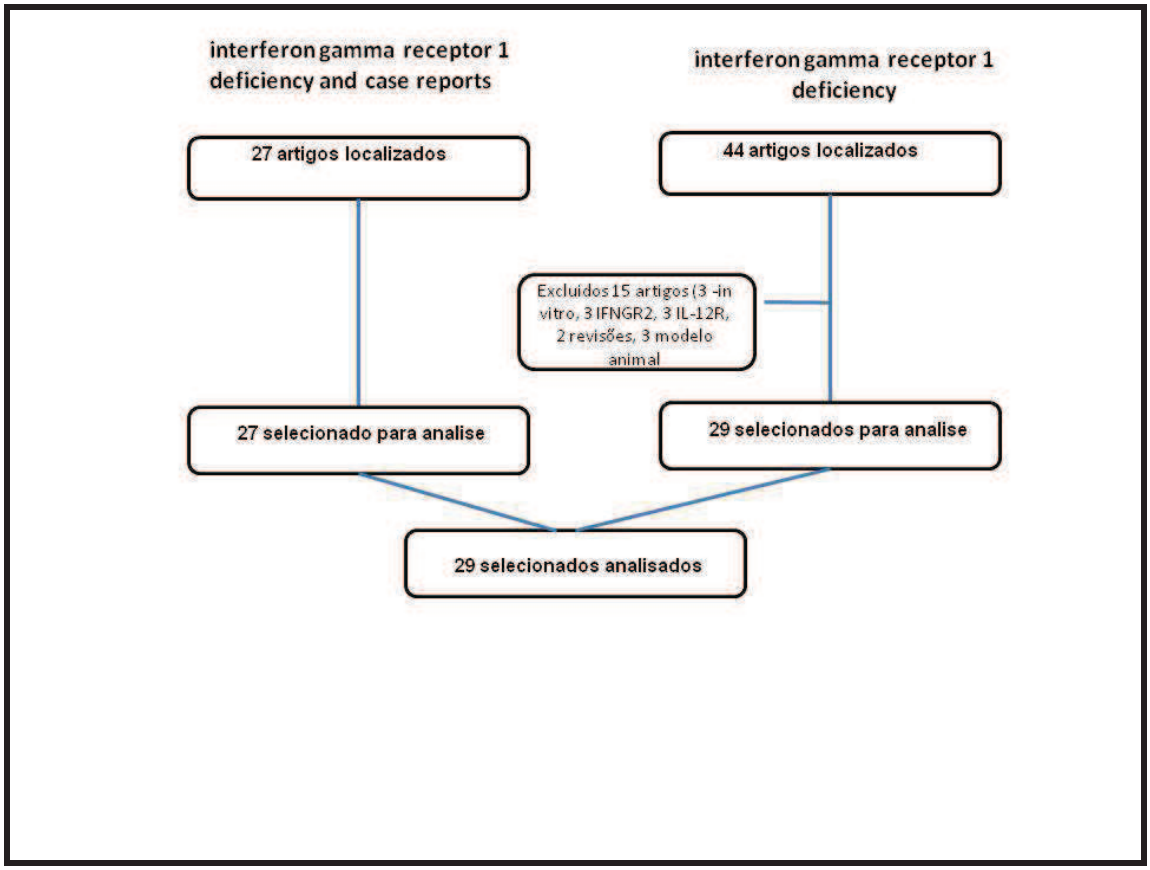
6.25) Deficiência do receptor 1 de IFN-γ

A busca realizada com o descritor ***"interferon gamma receptor 1 deficiency and case reports"*** **(Apêndice 71)** identificou 27 artigos, todos foram localizados na busca do item 25. Em nenhum destes relatos clínico houve menção a alterações orais.

Uma segunda busca foi realizada, mais aberta, aonde somente utilizamos o descritor ***"interferon gamma receptor 1 deficiency"*** **(Apêndice 72)** e restringimos para estudos murinos e revisões. Foram localizados 44 artigos e foram excluídos 15 artigos (3 estudos in vitro, 3 sobre IFNGR2, 3 sobre IL-12R, 2 revisões, 3 modelo animal). Foram incluídos para a análise 29 artigos sendo que 27 artigos já haviam sido 27 localizados na primeira busca. Em nenhum destes artigos houve relato clínico de manifestação oral **(Figura 25).**

Figura 25

Deficiência do receptor 1 de IFN-γ



6.26) Deficiência do receptor 2 de IFN-□

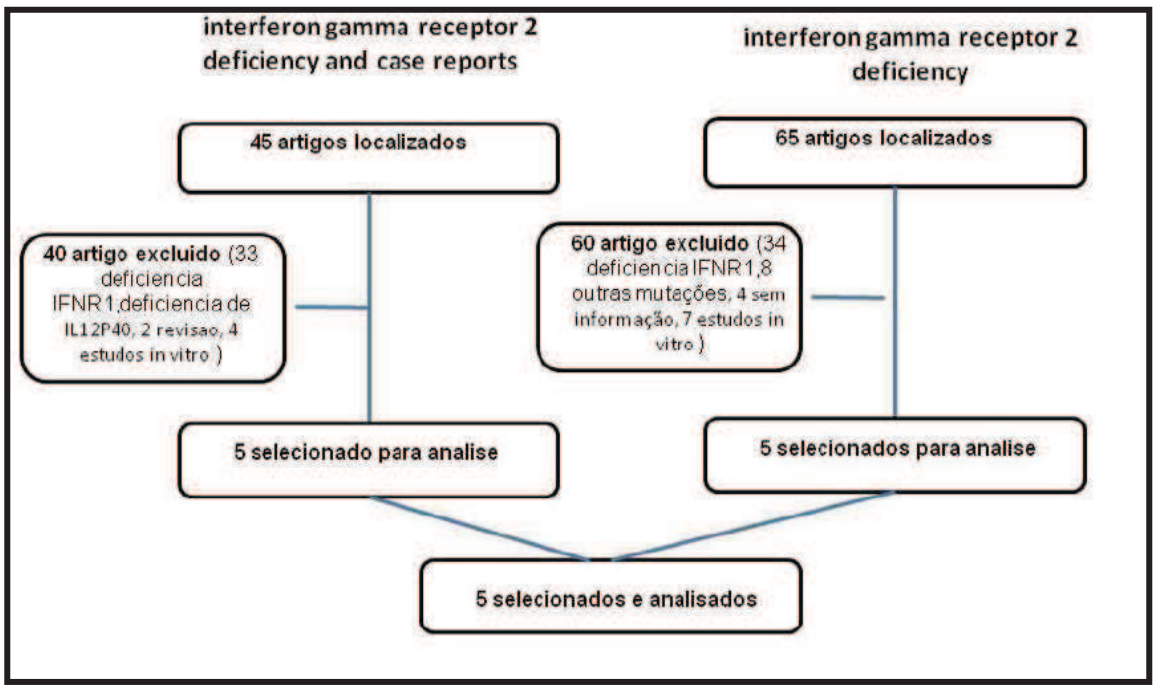
A busca realizada com o descritor ***"interferon gamma receptor 2 deficiency and case reports"*** **(Apêndice 73)** identificou 41 artigos e 33 artigos eram sobre pacientes com deficiência do receptor 1 de gama interferon, 1 artigo descreveu 2 pacientes sendo um com deficiência do receptor 1 e o outro do receptor 2, 1 artigo descrevia um paciente com deficiência de IL12P40 e somente 5 eram descrições de pacientes com deficiência do receptor 2. É importante ser mencionado que na maioria dos casos os

pacientes foram investigados para a deficiência do receptor 2. Nesses 5 artigos em que foram descritos 9 pacientes não houve relatos de alterações orais.

Uma segunda busca foi realizada, mais aberta, aonde somente utilizamos o descritor ***"interferon gamma receptor 2 deficiency"*** **(Apêndice 74)** e excluímos estudos murinos e revisões. Foram localizados 65 artigos e, como na busca anterior, 33 artigos eram de pacientes com mutações no receptor 1 de IFN-γ, 7 estudos *in vitro*, 3 em modelo animal, 2 artigos tinham pacientes com defeitos no receptor 1 e 2 de IFN-γ, 8 tratavam de pacientes com outro tipo de mutações, para 4 não obtivemos informações, 5 artigos eram sobre pacientes com mutação no receptor 2 de IFN- γ que já haviam sido localizados na primeira busca e que não continham nenhum relato clínico de alterações orais **(Figura 26).**

Figura 26

Deficiência do receptor 2 de IFN-γ

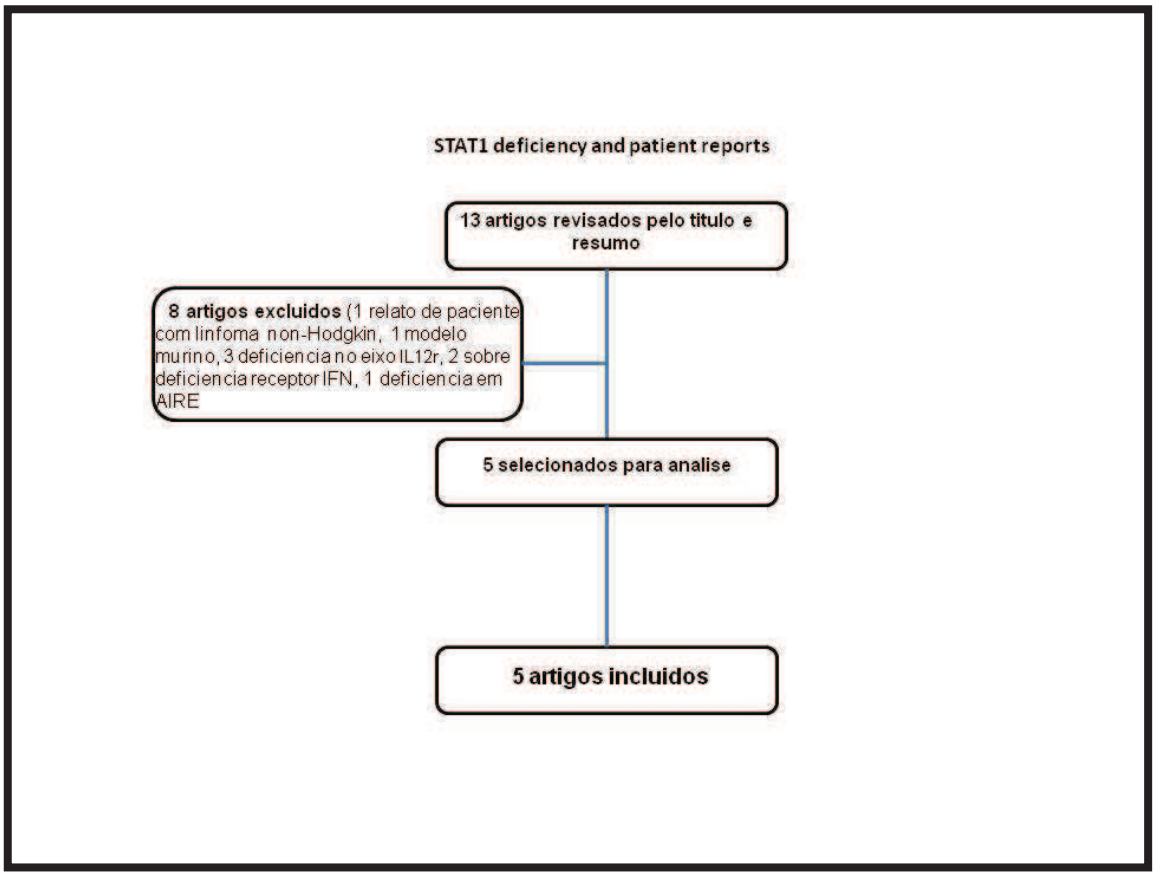


6.27) Deficiência de STAT1

A busca realizada utilizando ***"STAT1 deficiency and case reports"*** localizou 9 artigos **(Apêndice 75)**, e a segunda busca utilizando ***"STAT1 deficiency and patient reports"*** localizou 13 **(Apêndice 76)**, onde todos os artigos da primeira busca estavam incluídos. Foram excluídos 8 artigos por não relatarem deficiência de STAT1 **(Figura 27)** e foram analisados 5 artigos, totalizando 9 pacientes, nenhum dos quais relatado como tendo manifestações orais.

Figura 27

Deficiência de STAT1



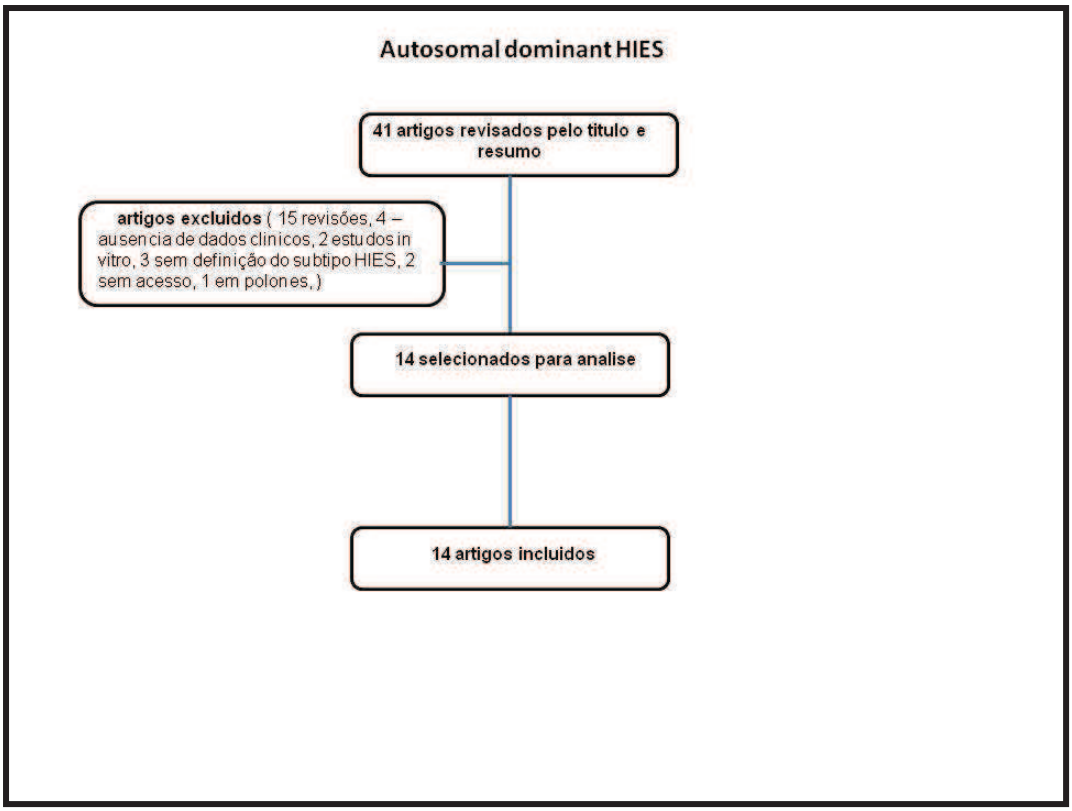
6.28) Síndrome de hiper IgE Autossômica Dominante

Foi realizada uma busca com ***"autosomal dominant hyper IgE and case report"*** **(Apêndice 77)** e foram localizados 8 artigos. Uma segunda ampliada utilizando ***"autosomal dominant hyper IgE"*** **(Apêndice 78)** localizou 41 artigos, incorporando todos da primeira busca.

A seleção dos artigos se baseou nos artigos que definiram a *AD hyper-IgE* e caracterizam o envolvimento de STAT3. É preciso ressaltar que na referência que utilizamos como base está sendo indicando o gene STAT1, mas toda a literatura recuperada relaciona a síndrome de hiper IgE autossômica dominante com STAT3 e não STAT1. Foram excluídos 27 artigos (15 revisões, 4 por ausência de dados clínicos, 2 estudos *in vitro*, 3 sem definição do subtipo HIES, 2 sem acesso, 1 em polonês) **(Figura 28).** Foram relatados 178 pacientes e 40,4% não apresentaram nenhuma manifestação oral. Dos 59,6% que apresentaram manifestações orais, 67,9% apresentaram retenção prolongada de dentes decíduos, 27,4% apresentaram candidíase oral e 3,8% relato de infecções dentárias e condições precárias dos dentes.

Figura 28

AD hyper-IgE



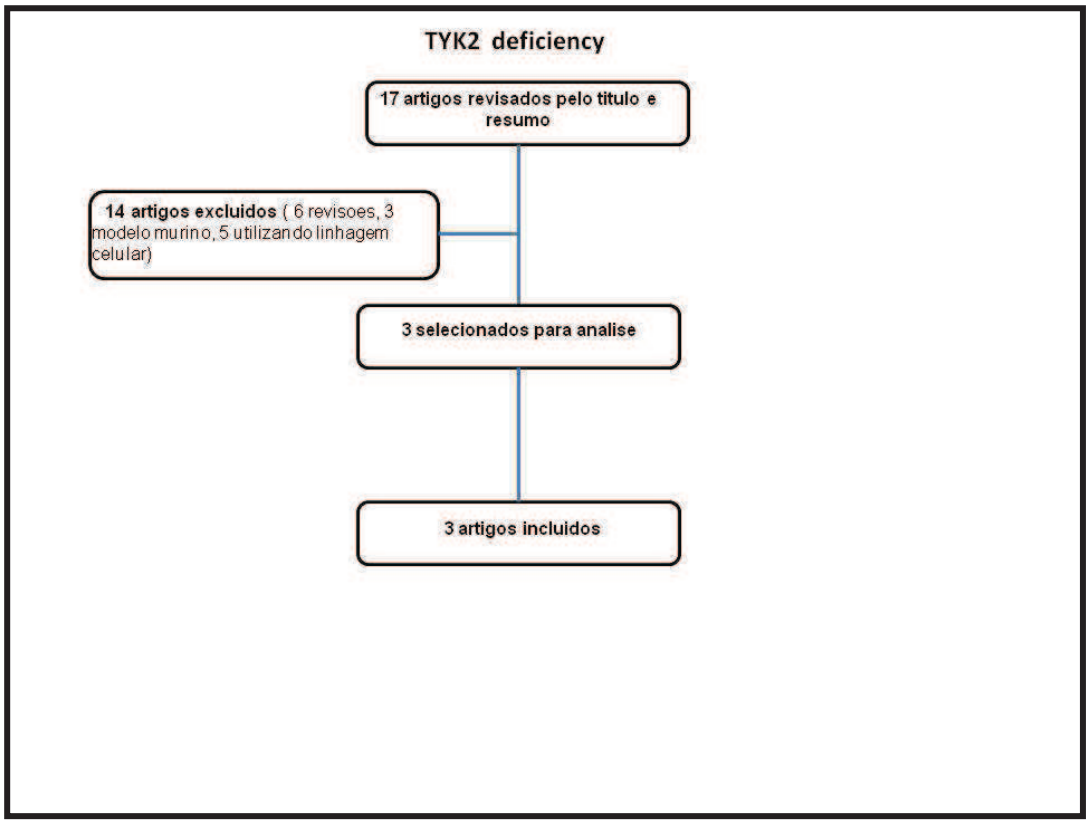
6.29) Síndrome de Hiper IgE autossômica recessiva

a) Deficiência de TYK2

A definição do descritor teve que ser feita utilizando *TYK2 kinase*. A busca de *"**TYK2 kinase deficiency and case reports**"* não encontrou nenhuma publicação. Passamos então para a busca combinando ***"TYK2 Kinase"[Mesh] AND deficiency and case reports"*** que localizou um relato de caso em 2006. Uma nova busca foi realizada utilizando ***"TYK2 Kinase"[Mesh] AND deficiency"*** e foram encontrados 34 artigos que, ao colocarmos o limite de busca para humanos, se reduziram a 17 artigos. **(Apêndice 79)**. Destes, 14

foram excluídos como mostrado na **Figura 29**. Dos 3 artigos analisados, correspondendo a 3 pacientes, 1 deles apresentou candidíase oral.

Figura 29
Deficiência de TYK2



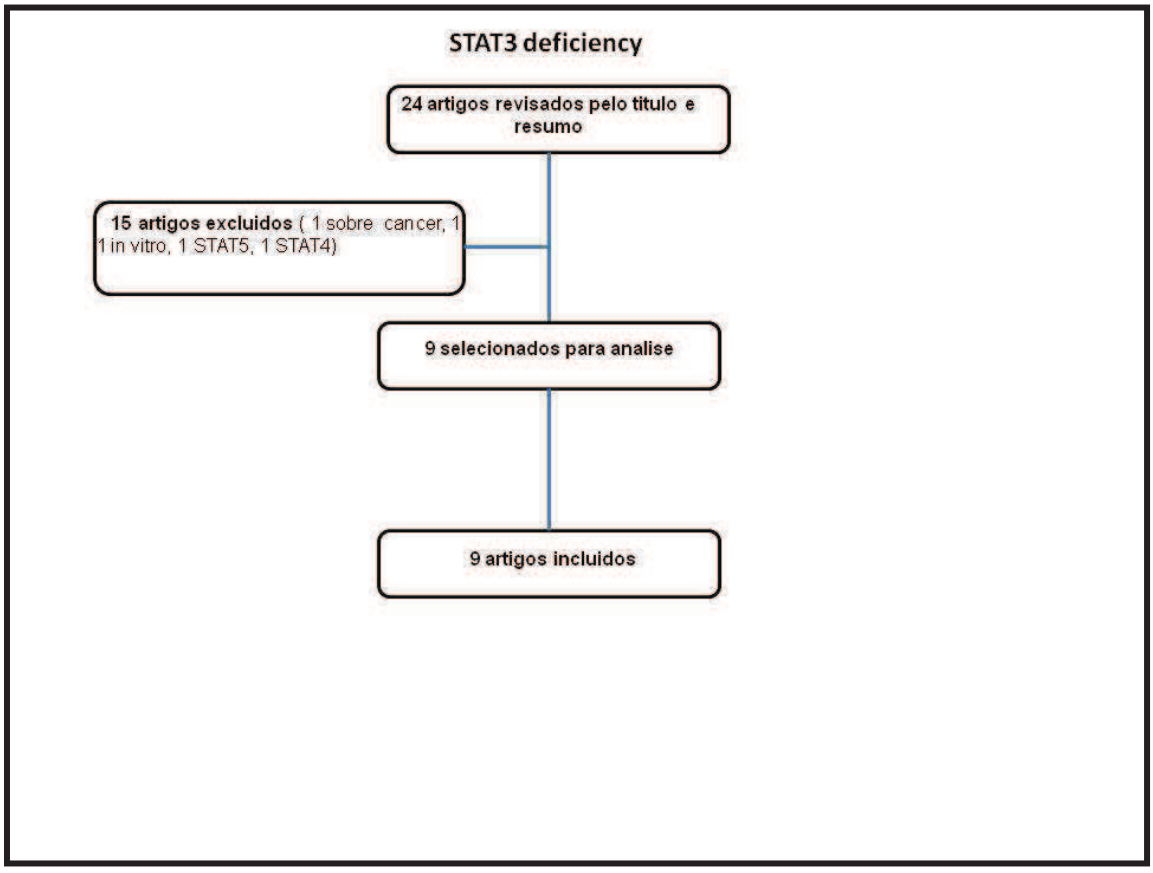
b) Deficiência de STAT3

A busca realizada utilizando *"**STAT3 Transcription Factor"[Mesh]) AND "Case Reports"*** com limite de busca em humanos em localizou 24 artigos **(Apêndice 80)** e 15 foram excluídos (10 sobre câncer, 1 em alemão, 1 em polonês, 1 sobre STAT5, 1 sobre STAT4, 1 estudo *in vitro*). Um total de 9 artigos foram selecionados **(Figura 30),** totalizando 19 pacientes analisados.

73,7% (n=14) dos pacientes não apresentavam manifestações orais e, dos 26,3% (n= 5 pacientes) apresentavam manifestações orais, 2 pacientes apresentavam com candidíase oral e 3 pacientes com retenção prolongada de dentes decíduos.

Figura 30

Deficiência de STAT3



6.30) Proteinose Alveolar Pulmonar

Iniciamos a busca utilizando ***“"Pulmonary alveolar proteinosis and case reports”*** e, limitando a estudos humanos e excluindo estudos em camundongo e revisões, localizamos 573 artigos. Refinamos a busca

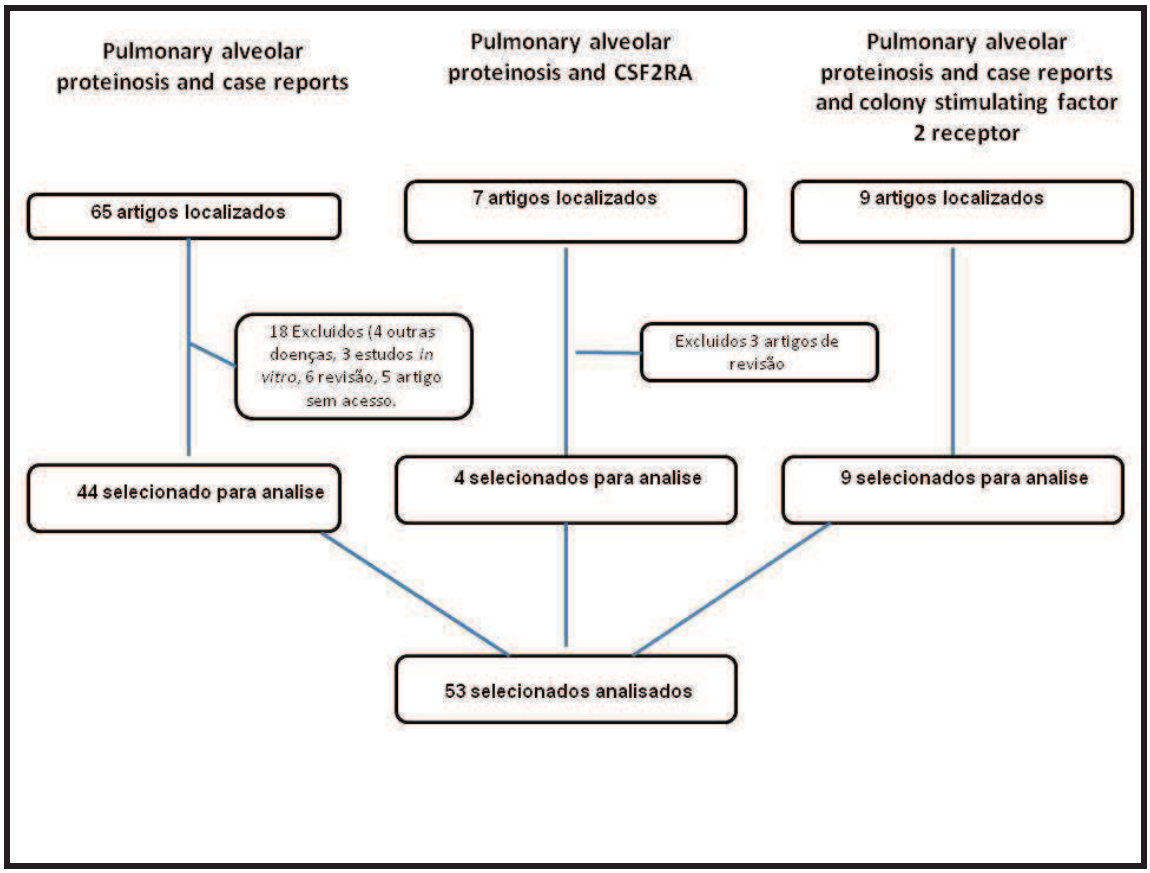
restringindo-nos aos artigos que associassem com **GM-CSF;** nessa seleção foram encontrados 62 artigos **(Apêndice 81)**, dos quais 4 não estavam relacionados com o assunto, 3 eram estudos *in vitro*, 6 eram artigos de revisão, 5 artigos aos quais não tivemos acesso. Foram analisados 44 artigos que totalizavam 53 pacientes, e não houve relato de manifestações orais em nenhum destes pacientes. É necessário mencionar que os relatos estavam muito direcionados para o diagnóstico da proteinose e dos aspectos dos vários tratamentos.

Uma segunda busca foi realizada utilizando os descritores ***"Pulmonary alveolar proteinosis and CSF2RA"*** **(Apêndice 82)** para relacionar com o gene identificado na classificação de IDP (Notarangelo, 2009) **(anexo 1).** Nessa busca encontramos 7 artigos, todos já localizados na primeira busca, 3 eram revisões e 4 relatos de casos. Destes relatos de casos, que totalizavam 11 pacientes a serem analisados, não houve nenhum relato de alterações orais.

O mesmo ocorreu na terceira busca ***"Pulmonary alveolar proteinosis and case reports and colony stimulating factor 2 receptor"*** **(Apêndice 83)** aonde foram encontrados 9 artigos, nenhum em sobreposição com a busca anterior, totalizando uma análise de 15 pacientes e nenhum relato de manifestação oral **(Figura 31).**

Figura 31

Proteinose Alveolar Pulmonar



6.31) Análise global e comparativa dos resultados:

A revisão sistemática da literatura científica, realizada para avaliar a freqüência das manifestações orais e dentárias nas IDP com alterações de número e/ou função de fagócitos permitiu analisar dados de um total de 1721 pacientes, relatados nos 632 artigos analisados.

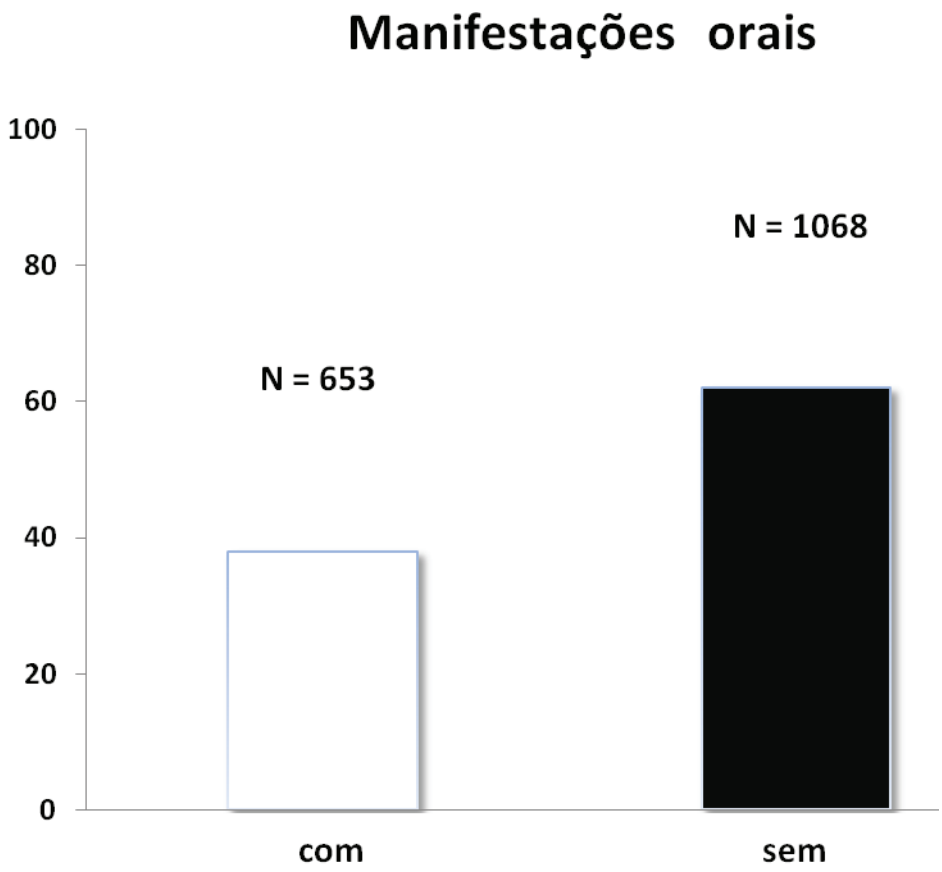
Dentre estes 1721 pacientes, 653 (37,9%) apresentaram relato de alguma manifestação oral e/ou dentária na descrição clínica do caso, enquanto 1068 não apresentaram qualquer relato de manifestação oral e/ou dentária. A

Figura 31 mostra o percentual de pacientes quanto à presença ou ausência de relatos de manifestações orais e dentárias nas descrições dos casos clínicos dos artigos analisados.

Os dados dos resultados encontrados na revisão sistemática estão sintetizados nas tabelas 1 e 2. A **Tabela 1** apresenta os dados das freqüências das manifestações orais e dentárias relatadas nos pacientes analisados, de acordo com a ordem seqüencial proposta na classificação de IDP (Notarangelo, 2009) **(Anexo 1)**.

Já a **Tabela 2** apresenta a síntese das manifestações orais e dentárias relatadas nas desordens de fagócitos que apresentaram, em algum momento da análise, relato de manifestação oral e dentária, ou seja: Neutropenias Congênitas Graves; Síndrome de Kostmann; Neutropenias com malformações cardíacas e urogenitais; Neutropenia Cíclica; LAD-1; LAD-2; LAD-3; Periodontite Juvenil Localizada; Síndrome de Papillon-Lefrèvre; Síndrome de Schwachman-Diamond; Doença Granulomatosa Crônica ligada ao X; Doença Granulomatosa Crônica autossômica; Deficiência da cadeia β-1 do receptor de IL-12 e IL-23; Deficiência de STAT-1; Síndrome de Hiper IgE autossômica dominante e Síndrome de Hiper IgE autossômica recessiva.

Figura 31



	1) Neutropenias Congênitas Grave (ELA2)		2) Neutropenias Congênitas Grave (GFI1)		3) Kostmann		4) Neutropenia com malformações cardiacas e urogenitais		5) Glycogen storage disease type 1b		6) Neutropenia Cíclica.		7) Neutropenia ligada ao X/ mielodisplasia		8) Deficiencia de P14		9) deficiencia de Adesão leucocitário do tipo I	
total de pacientes	20		4		77		15		143		46		41		1		80	
pacientes com alterções orais	3	15%	0	0%	44	57%	4	27%	0	0%	34	74%	0	0%	0	0%	15	18,8%
		%				%		%										
Periodontite	1	33,3	0	0	30	68,2	2	50,00	0	0	5	14,7	0	0	0	0	8	53,3
Perda precoce de dentes decíduos:	0	0	0	0	2	4,5	3	75,00	0	0	2	5,9	0	0	0	0	0	0
Gengivite:	1	33,3	0	0	13	29,5	1	25,00	0	0	4	11,8	0	0	0	0	5	33,3
Aftas:ulceras aftosas	2	66,7	0	0	27	61,4	3	75,00	0	0	14	41,2	0	0	0	0	1	6,7
Candidíase:	0	0	0	0	9	20,5	0	0	0	0	0	0	0	0	0	0	2	13,3
Perda de osso alveolar	0	0	0	0	2	4,5	0	0,00	0	0	0	0	0	0	0	0	2	13,3
perda dentaria	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
Mobilidade dentária:	0	0	0	0	2	4,5	0	0	0	0	0	0	0	0	0	0	2	13,3
Estomatite	1	33,3	0	0	3	6,8	0	0	0	0	6	17,6	0	0	0	0	2	13,3
Envolvimento oral	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
dentinogenesis imperfecta	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
Abscesso de glândula parótida:	0	0	0	0	1	2,3	0	0	0	0	0	0	0	0	0	0	0	0
abcesso	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
Infecções orais de repetição	0	0	0	0	1	2,3	0	0	0	0	0	0	0	0	0	0	0	0
Odontochondrodysplasia	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
carie	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
Infecções orais por herpes	0	0	0	0	1	0,0	0	0	0	0	0	0	0	0	0	0		
absorção de mandibula e maxilar	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
paralisia do nervo facial	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
Retenção prolongada dos dentes decíduos	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
retração gengiva	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0

10) deficiencia de Adesão leucocitário do tipo II		11) deficiencia de Adesão leucocitário do tipo III		12) Rac2 deficiency		13) deficiencia de actina b		14) periodontite juvenil localizada - FPR1: Chemokine		15) Síndrome de Papillon-Lefèvre		16) Deficiencia de granulo especifica (proteina CEBPE)		17) Shwachman-Diamond syndrome		18) Doença granulomatosa crônica ligada ao X (CGD)		19) Doença granulomatosa crônica autossômica (CGD) - CYBA		20) Doença granulomatosa crônica autossômica (CGD) - NCF1:		21) Doença granulomatosa crônica autossômica (CGD) – NCF2		22) Deficiência da cadeia b1 do receptor de IL-12 and IL-23	
21		52		4		3		223		152		6		131		89		11		46		17		55	
10	48%	28	54%	0	0%	0	0%	223	100%	140	92%	0	0%	15	11%	12	13,5%	0	0%	7	15%	1	6%	9	16%
100	100	2	7,1	0	0	0	0	223	100,0	75	53,6	0	0	0	0	0	0	0	0	0	0	0	0	0	0
0	0	1	3,6	0	0	0	0	0	0	60	42,9	0	0	2	13,3	0	0	0	0	0	0	0	0	0	0
0	0	4	14,3	0	0	0	0	0	0	44	31,4	0	0	0	0	0	0	0	0	0	0	0	0	0	0
0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	4	33,3	0	0	1	14,3	0	0	0	0
0	0	3	10,7	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	1	6,0	4	44,4
0	0	0	0	0	0	0	0	0	0	25	17,9	0	0	0	0	0	0	0	0	0	0	0	0	0	0
0	0	0	0	0	0	0	0	0	0	29	20,7	0	0	0	0	0	0	0	0	0	0	0	0	0	0
0	0	0	0	0	0	0	0	0	0	24	17,1	0	0	0	0	0	0	0	0	0	0	0	0	0	0
0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	8	66,7	0	0	1	14,3	0	0	3	33,3
0	0	8	28,6	0	0	0	0	0	0	0	0	0	0	1	6,7	0	0	0	0	0	0	0	0	0	0
0	0	0	0	0	0	0	0	0	0	1	0,7	0	0	6	0	0	0	0	0	0	0	0	0	0	0
0	0	0	0	0	0	0	0	0	0			0	0	0	0	0	0	0	0	0	0	0	0	0	0
0	0	0	0	0	0	0	0	0	0	6	4,3	0	0	0	0	0	0	0	0	0	0	0	0	0	0
0	0	0	0	0	0	0	0	0	0			0	0	0	0	0	0	0	0	0	0	0	0	0	0
0	0	0	0	0	0	0	0	0	0	0	0	0	0	3	20,0	0	0	0	0	0	0	0	0	0	0
0	0	0	0	0	0	0	0	0	0	3	2,1	0	0	0	0	0	0	0	0	0	0	0	0	0	0
		0	0	0	0	0	0	0	0			0	0	0	0	0	0	0	0	0	0	0	0	1	11
0	0	0	0	0	0	0	0	0	0	2	1,4	0	0	0	0	0	0	0	0	0	0	0	0	0	0
0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	1	11,1
1	10,0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
0	0	0	0	0	0	0	0	0	0	1	0,7	0	0	0	0	0	0	0	0	0	0	0	0	0	0

23) Deficiência de IL-12p40		24) Deficiência do receptor 1 de IFN-γ		25) Deficiência do receptor 2 de IFN-γ		26) STAT 1		27) AD hyper-IgE (STAT3 deficiency)		28) AR hyper-IgE (TYK2 deficiency)		28) AR hyper-IgE (STAT3 deficiency)		29) Proteinose Alveolar Pulmonar		total	
18		134		65		9		178		3		9		68		1721	
0	0%	0	0%	0	0%	1	11%	106	60%	1	33%	0	0%	0	0%	653	38%
0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0		
0	0	0	0	0	0	0	0	72	67,9	0	0	0	0	0	0		
0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0		
0	0	0	0	0	0	1	2	0	0	0	0	0	0	0	0		
0	0	0	0	0	0	0	0	29	27,4	1	33,0	0	0	0	0		
0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0		
0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0		
0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0		
0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0		
0	0	0	0	0	0	0	0	1	0,9	0	0	0	0	0	0		
0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0		
0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0		
0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0		
0	0	0	0	0	0	0	0	4	3,8	0	0	0	0	0	0		
0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0		
0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0		
0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0		
0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0		
0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0		
0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0		
0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0		

TABELA 2

	1) Neutropenias Congênitas Grave (ELA2)	3) Kostmann	4) Neutropenia com malformações cardiacas e urogenitais	6) Neutropenia Cíclica.	9) deficiencia de Adesão leucocitário do tipo I	10) deficiencia de Adesão leucocitário do tipo II	11) deficiencia de Adesão leucocitário do tipo III	14) periodontite juvenil localizada - FPR1: Chemokine receptor	15) Síndrome de Papillon-Lefèvre
total de pacientes	20	77	15	46	80	21	52	223	152
pacientes com alterções orais	3	44	4	34	15	10	28	223	140
%	15,0%	57,1%	26,7%	73,9%	18,8%	47,6%	53,8%	100,0%	92,1%
N=									
Periodontite	1	30	2	5	8	10	2	223	75
Perda precoce de dentes decíduos	0	2	3	2	0	0	1	0	60
Gengivite:	1	13	1	4	5	0	4	0	44
Aftas:ulceras aftosas	2	27	3	14	1	0	0	0	0
Candidíase:	0	9	0	0	2	0	3	0	0
Perda de osso alveolar	0	2	0	0	2	0	0	0	25
perda dentaria	0	0	0	0	0	0	0	0	29
Mobilidade dentária	0	2	0	0	2	0	0	0	24
Estomatite	1	3	0	6	2	0	0	0	0
Envolvimento oral	0	0	0	0	0	0	8	0	0
dentinogenesis imperfecta	0	0	0	0	0	0	0	0	1
Abscesso de glândula parótida	0	1	0	0	0	0	0	0	0
abcesso	0	0	0	0	0	0	0	0	6
Infecções orais de repetição	0	1	0	0	0	0	0	0	
Odontochondrodysplasia	0	0	0	0	0	0	0	0	0
carie	0	0	0	0	0	0	0	0	3
Infecções orais por herpes	0	1	0	0	0	0	0	0	0
absorção de mandibula e maxilar	0	0	0	0	0	0	0	0	2
paralisia do nervo facial	0	0	0	0	0	0	0	0	0
Retenção prolongada dos dentes decíduos	0	0	0	0	0	1	0	0	0
retração gengiva	0	0	0	0	0	0	0	0	1

17) Shwachman-Diamond syndrome	18) Doença granulomatosa crônica ligada ao X (CGD)	20) Doença granulomatosa crônica autossômica (CGD) - NCF1:	21) Doença granulomatosa crônica autossômica (CGD) – NCF2	22) Deficiência da cadeia b1 do receptor de IL-12 and IL-23	26) STAT 1	27) AD hyper-IgE (STAT3 deficiency)	28) AR hyper-IgE (TYK2 deficiency)	TOTAL
131	89	46	17	55	9	178	3	1194
15	12	7	1	9	1	106	1	653
11,5%	13,5%	15,2%	5,9%	16,4%	11,1%	59,6%	33,3%	54,7%
0	0	0	0	0	0	0	0	
2	0	0	0	0	0	72	0	
0	0	0	0	0	0	0	0	
0	4	1	0	0	1	0	0	
0	0	0	1	4	0	29	1	
0	0	0	0	0	0	0	0	
0	0	0	0	0	0	0	0	
0	0	0	0	0	0	0	0	
0	8	1	0	3	0	0	0	
1	0	0	0	0	0	1	0	
6	0	0	0	0	0	0	0	
0	0	0	0	0	0	0	0	
0	0	0	0	0	0	0	0	
0	0	0	0	0	0	4	0	
3	0	0	0	0	0	0	0	
0	0	0	0	0	0	0	0	
0	0	0	0	1	0	0	0	
0	0	0	0	0	0	0	0	
0	0	0	0	1	0	0	0	
0	0	0	0	0	0	0	0	
0	0	0	0	0	0	0	0	

Analisamos também a distribuição dos pacientes estudados de acordo com o tipo de desordens de número e/ou função de fagócitos analisada. (**Figura 32)**. De uma maneira geral, foi analisado um número significativo de casos relatados pelo tipo de doença de fagócitos. Algumas doenças estudadas, porém, possuem poucos relatos de caso clínicos descritos na literatura, principalmente as mais recentemente reconhecidas, pois nestes casos ocorre uma maior ênfase na caracterização molecular da doença.

Outro aspecto analisado foi a comparação entre a freqüência das diferentes manifestações orais relatadas nos casos clínicos analisados **(Figura 33).** Nessa análise encontramos de uma maneira impactante, um total de 68% de doença periodontal, correspondente a 446 pacientes dos 653 pacientes com manifestação oral e dentária. A doença periodontal foi, portanto, a principal manifestação oral relatada nos pacientes com defeitos de fagócitos. A segunda manifestação oral mais relatada dentre os casos analisados foi a perda precoce de dentes decíduos, encontrada em 142 pacientes (21,7%). A terceira manifestação mais relatada foi a gengivite, encontrada em 72 pacientes (11,0%). Cabe ressaltar que nos artigos que relatavam sangramento gengival, este foi entendido como inerente da doença de gengivite e consolidados em uma mesma manifestação. O termo gengivite apareceu em 54 pacientes e o termo sangramento gengival em 18 pacientes. A quarta manifestação mais relatada foram as aftas, encontradas em 53 pacientes (8,1%). A Candidíase oral foi encontrada em 7,5% dos casos (n=49). Em seguida, a perda de osso alveolar e a perda dentária, encontradas em 4,4% (n=29) e a mobilidade dentária, encontrada em 28 pacientes (4,3%). O termo estomatite foi relatado em 24 pacientes (3,7%). Algumas manifestações estiveram presente entre 10 a

5 pacientes: O termo inespecífico envolvimento da saúde oral foi relato em 1,5% dos casos (n=10); a dentinogênese imperfecta em 1,1% dos casos (n=7); os abscessos em 0,9% dos casos (n=6); o termo inespecífico infecções orais de repetição em 0,8% (n=5). Algumas manifestações estiveram presentes em 1 a 3 pacientes: odontocondrodisplasia em 0,5% (n=3); a doença cárie em 0,5% (n=3), as infecções orais por herpes em 0,3% (n=2); a reabsorção óssea de mandíbula e maxila em 0,3% (n=2); o abscesso de glândula parótida em 0,2% (n=1); a paralisia do nervo facial em 0,2% (n=1), a retenção prolongada de dentes decíduos em 0,2% (n=1) e a retração gengiva 0,2% (n=1)

Outra análise realizada comparou as diferentes manifestações orais e dentárias com o tipo de célula primariamente afetado, baseando-se na classificação de IDP (Notarangelo, 2009) **(Anexo 1)**.

Figura 32

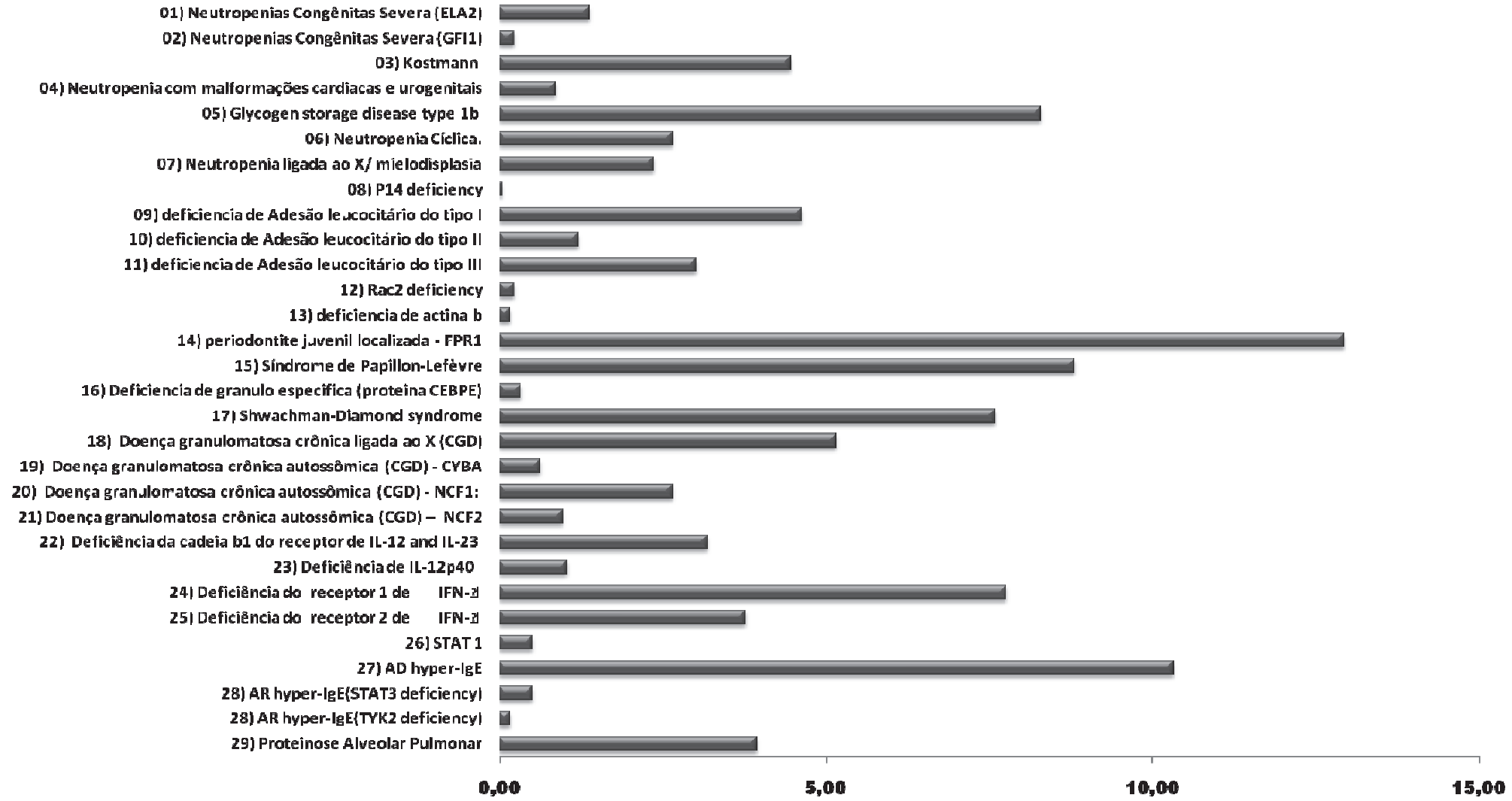

01) Neutropenias Congênitas Severa (ELA2)
02) Neutropenias Congênitas Severa (GFI1)
03) Kostmann
04) Neutropenia com malformações cardiacas e urogenitais
05) Glycogen storage disease type 1b
06) Neutropenia Cíclica.
07) Neutropenia ligada ao X/ mielodisplasia
08) P14 deficiency
09) deficiencia de Adesão leucocitário do tipo I
10) deficiencia de Adesão leucocitário do tipo II
11) deficiencia de Adesão leucocitário do tipo III
12) Rac2 deficiency
13) deficiencia de actina b
14) periodontite juvenil localizada - FPR1
15) Síndrome de Papillon-Lefèvre
16) Deficiencia de granulo especifica (proteina CEBPE)
17) Shwachman-Diamond syndrome
18) Doença granulomatosa crônica ligada ao X (CGD)
19) Doença granulomatosa crônica autossômica (CGD) - CYBA
20) Doença granulomatosa crônica autossômica (CGD) - NCF1:
21) Doença granulomatosa crônica autossômica (CGD) – NCF2
22) Deficiência da cadeia b1 do receptor de IL-12 and IL-23
23) Deficiência de IL-12p40
24) Deficiência do receptor 1 de IFN-
25) Deficiência do receptor 2 de IFN-
26) STAT 1
27) AD hyper-IgE
28) AR hyper-IgE(STAT3 deficiency)
28) AR hyper-IgE(TYK2 deficiency)
29) Proteinose Alveolar Pulmonar
0,00
5,00
10,00
15,00

Figura 33

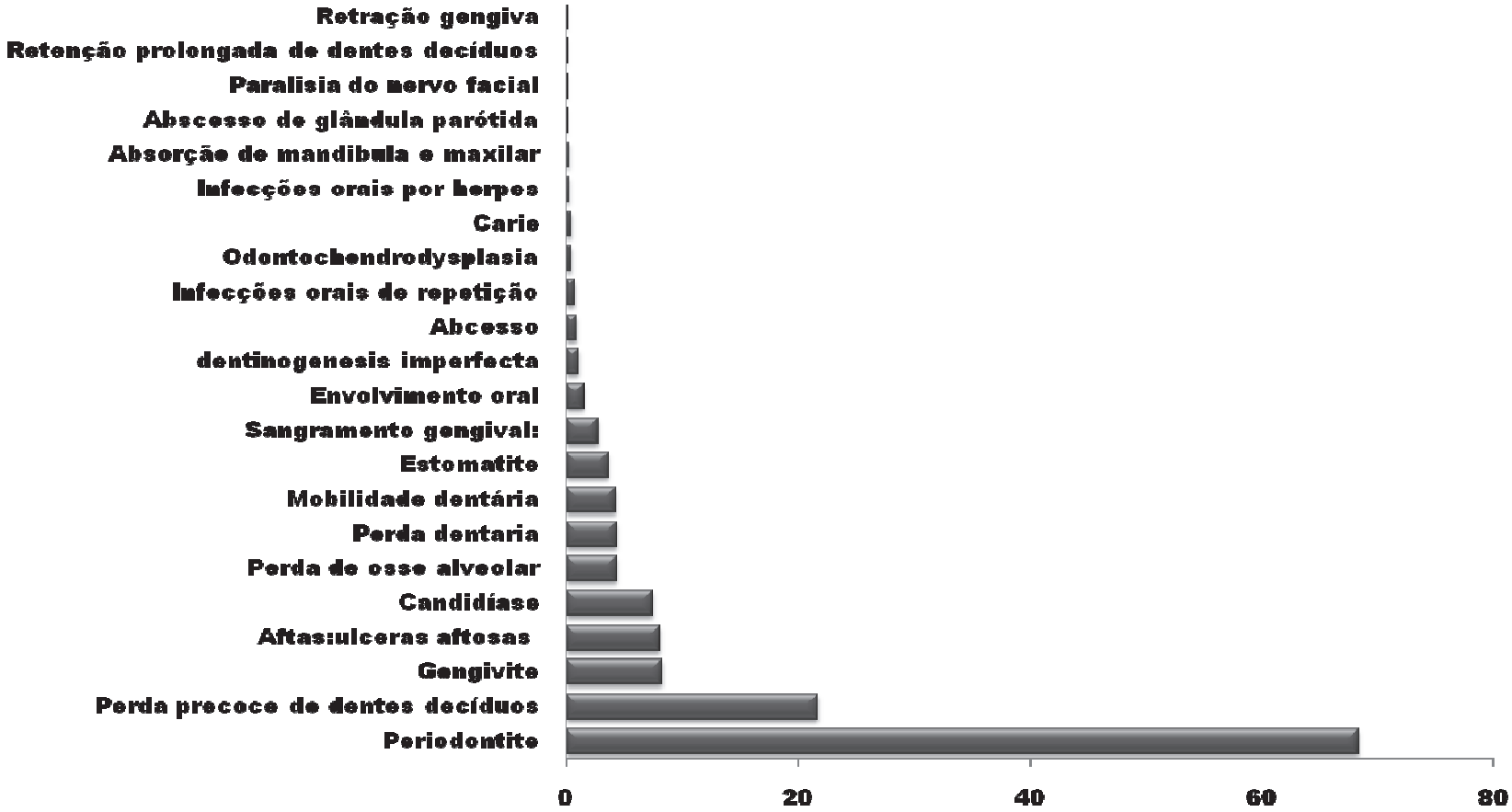

Retração gengiva
Retenção prolongada de dentes decíduos
Paralisia do nervo facial
Abscesso de glândula parótida
Absorção de mandibula e maxilar
Infecções orais por herpes
Carie
Odontochondrodysplasia
Infecções orais de repetição
Abcesso
dentinogenesis imperfecta
Envolvimento oral
Sangramento gengival:
Estomatite
Mobilidade dentária
Perda dentaria
Perda de osse alveolar
Candidíase
Aftas:ulceras aftosas
Gengivite
Perda precoce de dentes decíduos
Periodontite
0
20
40
60
80

Figura 34

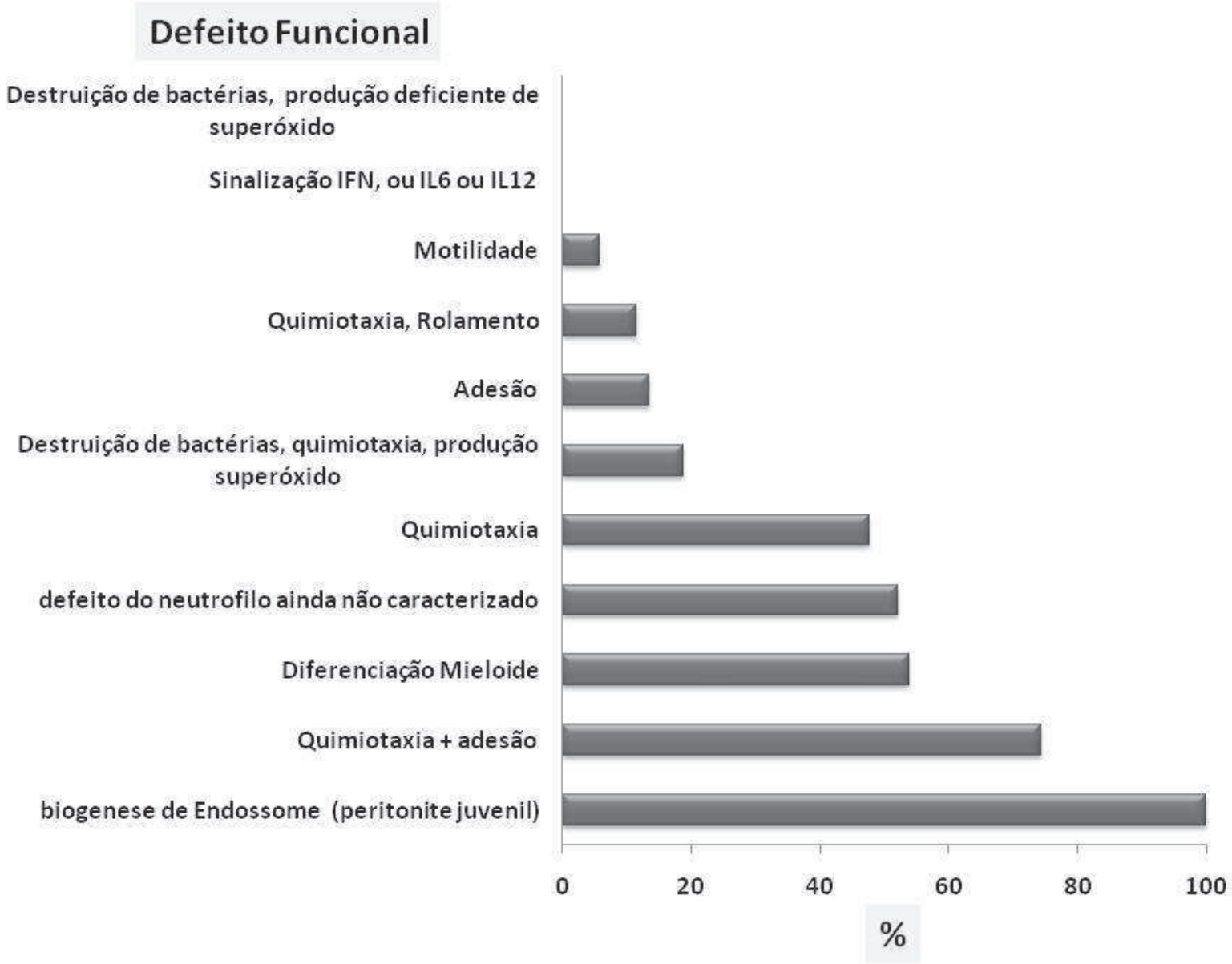
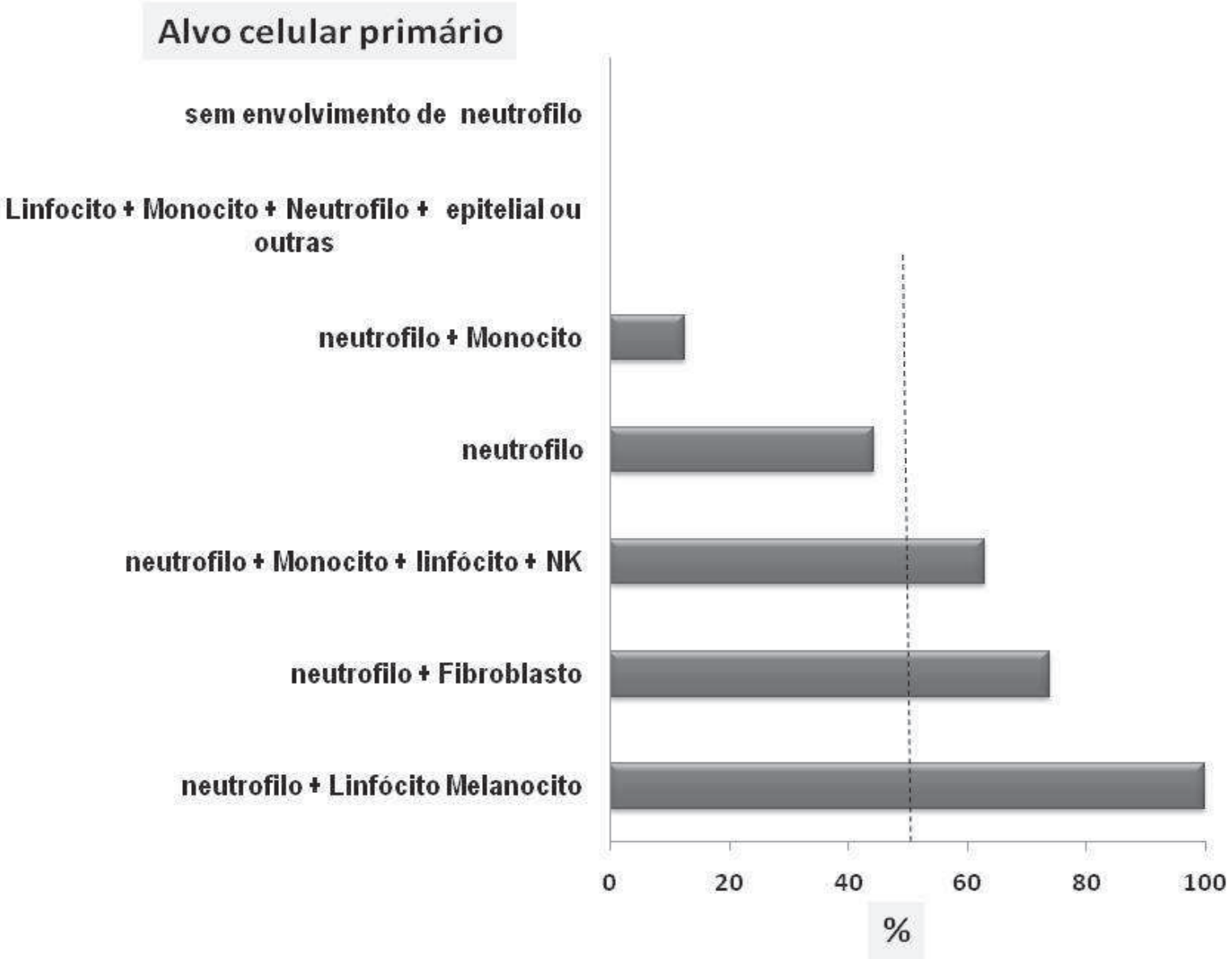

Defeito Funcional
Destruição de bactérias, produção deficiente de superóxido
Sinalização IFN, ou IL6 ou IL12
Motilidade
Quimiotaxia, Rolamento
Adesão
Destruição de bactérias, quimiotaxia, produção superóxido
Quimiotaxia
defeito do neutrofilo ainda não caracterizado
Diferenciação Mieloide
Quimiotaxia + adesão
biogenese de Endossome (peritonite juvenil)
0
20
40
60
80
100
%

Figura 35


Alvo celular primário
sem envolvimento de neutrofilo
Linfocito + Monocito + Neutrofilo + epitelial ou outras
neutrofilo + Monocito
neutrofilo
neutrofilo + Monocito + linfócito + NK
neutrofilo + Fibroblasto
neutrofilo + Linfócito Melanocito
0
20
40
60
80
100
%

6.32) Validação da estratégia inicial de busca

Após a realização de todas as buscas, comparamos os artigos se os artigos recuperados na busca inicial sobre deficiência de células fagocitárias estavam relacionados nas demais buscas realizadas. Da busca inicial, dos 124 selecionados para a análise, excluímos 87 artigos que não se tratavam de deficiência de fagócitos e analisamos se os 37 artigos sobre deficiência de número e/ou função de fagócitos localizados na busca inicial estavam também nas demais buscas realizadas (**Figura 36**)

A **Figura 37** apresenta um levantamento dos artigos localizados na primeira busca de acordo com os assuntos relacionados. Nota-se que a grande maioria dos artigos trata-se de Deficiência Seletiva de IgA, que é a imunodeficiência mais prevalente.

Dentro das IDP relacionadas a defeitos em número e/ou função de fagócitos, alvo deste trabalho, ao compararmos os artigos localizados na primeira busca com os artigos das demais buscas realizadas, foi possível observar que 100% dos artigos sobre Síndrome de Hiper IgE e Deficiência de Adesão Leucocitária encontrados no primeiro estudo também haviam sido inclusos para análise nas respectivas buscas. Em relação aos 50% de sobreposição para os artigos localizados para a Doença Granulomatosa Crônica, isto provavelmente deve-se ao fato de que nas buscas conseguintes, utilizamos os descritores propostos pela Classificação de IDP (Notarangelo, 2009), separando esta doença em autossômica ou ligada ao X e, como esta diferenciação etiológica ocorreu posteriormente, alguns artigos mais antigos não foram selecionados nestas buscas.

A **Figura 38** mostra que dentre os artigos localizados na primeira busca que induziu o nosso estudo, as doenças da Tabela V da Classificação de IDP (Notarangelo, 2009) (**Anexo 1**), correspondem a segunda maior categoria de estudos de manifestações orais e dentárias, ficando atrás apenas na Deficiências de IgA. Isto corrobora para a nossa idéia inicial de que as desordens de número e/ou função de fagócitos cursam com manifestações orais e dentárias e que o nosso objetivo de estudar a freqüência destas manifestações se deve a lacuna de estudos epidemiológicos.

FIGURA 36

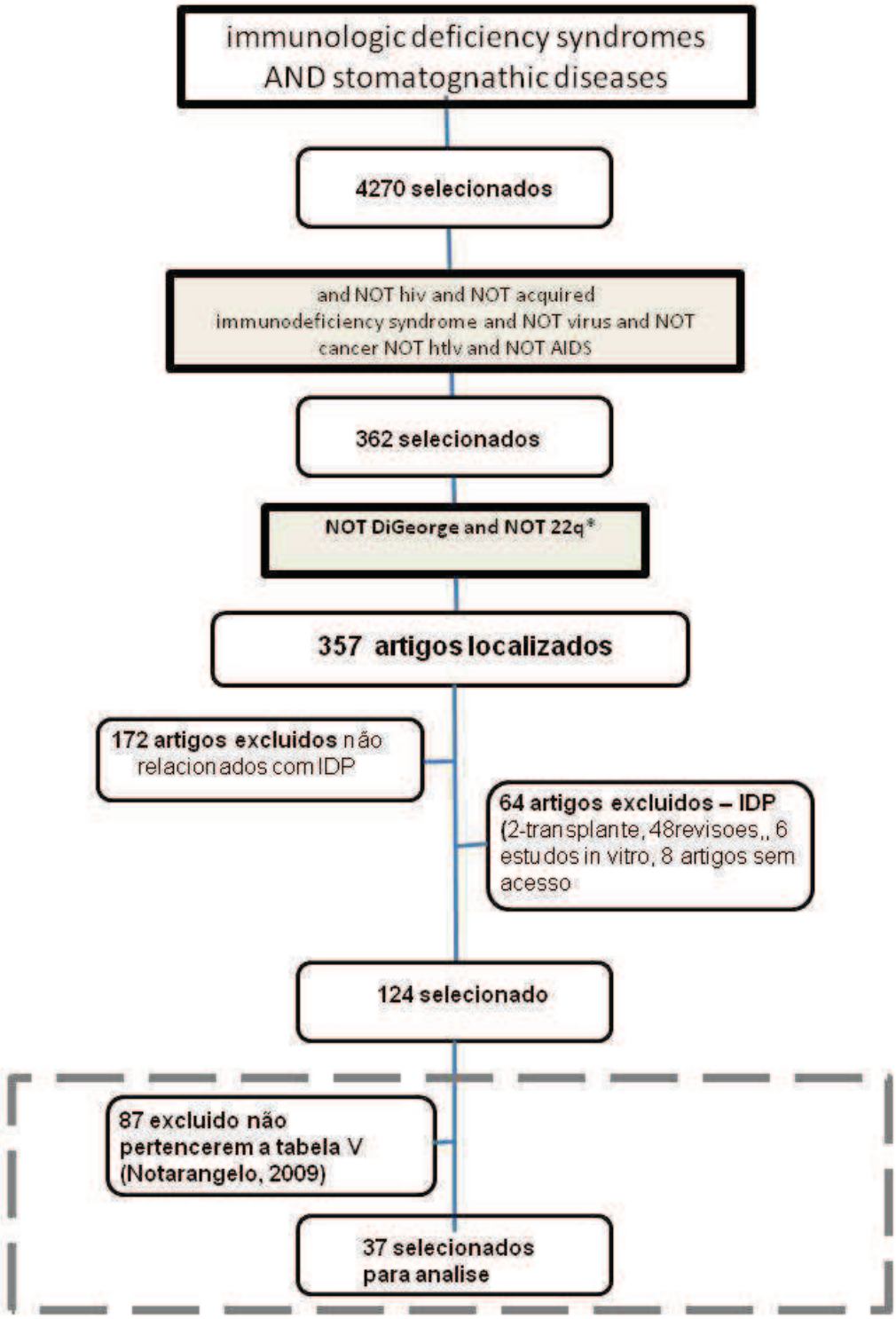

immunologic deficiency syndromes AND stomatognathic diseases
4270 selecionados
and NOT hiv and NOT acquired immunodeficiency syndrome and NOT virus and NOT cancer NOT htlv and NOT AIDS
362 selecionados
NOT DiGeorge and NOT 22q*
357 artigos localizados
172 artigos excluidos não relacionados com IDP
64 artigos excluidos – IDP (2-transplante, 48revisoes,, 6 estudos in vitro, 8 artigos sem acesso
124 selecionado
87 excluido não pertencerem a tabela V (Notarangelo, 2009)
37 selecionados para analise

FIGURA 37

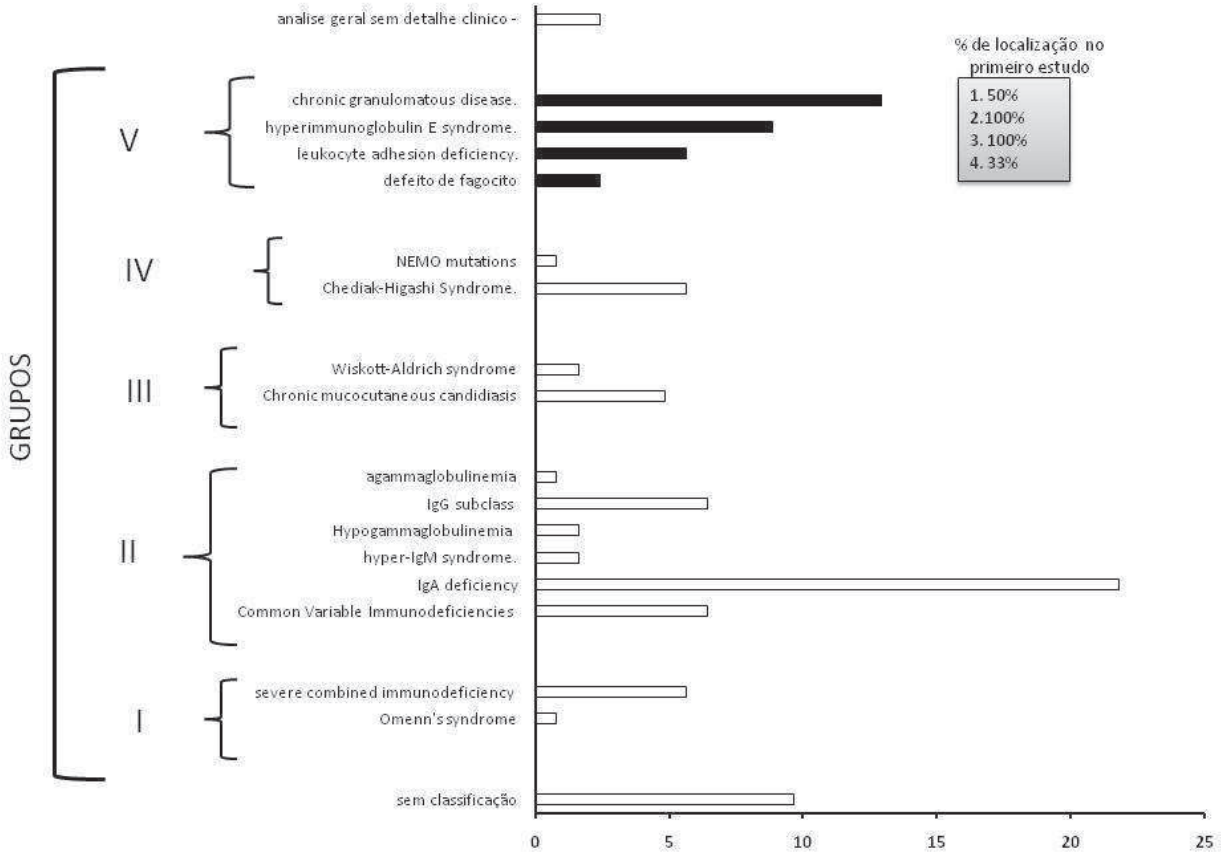
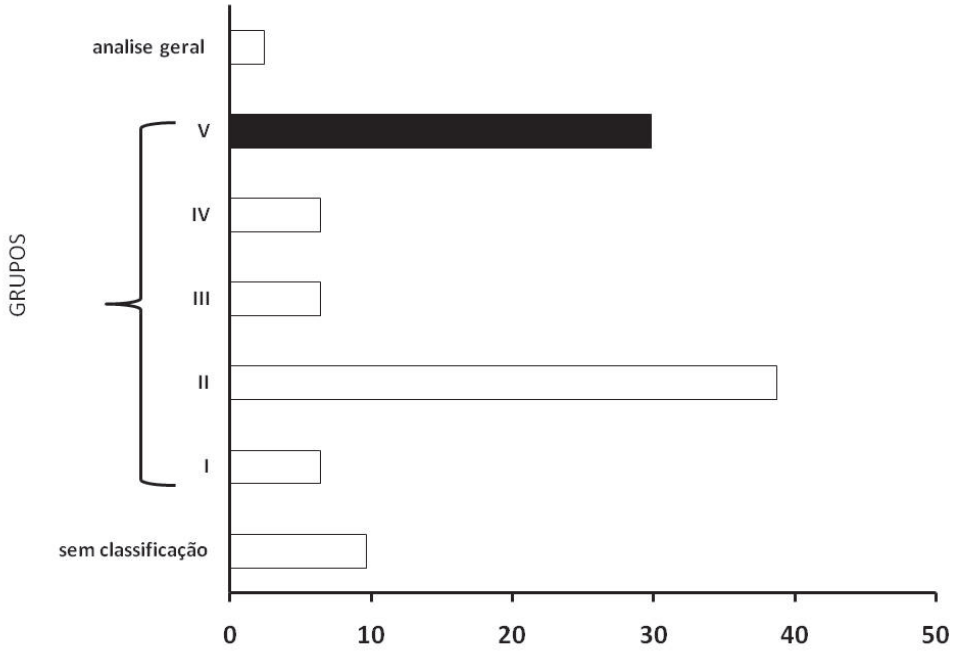

GRUPOS
V
IV
III
II
I
analise geral sem detalhe clinico -
chronic granulomatous disease.
hyperimmunoglobulin E syndrome.
leukocyte adhesion deficiency.
defeito de fagocito
NEMO mutations
Chediak-Higashi Syndrome.
Wiskott-Aldrich syndrome
Chronic mucocutaneous candidiasis
agammaglobulinemia
IgG subclass
Hypogammaglobulinemia
hyper-IgM syndrome.
IgA deficiency
Common Variable Immunodeficiencies
severe combined immunodeficiency
Omenn's syndrome
sem classificação
% de localização no primeiro estudo
1. 50%
2.100%
3. 100%
4. 33%
0
5
10
15
20
25

FIGURA 38


analise geral
V
IV
III
II
I
sem classificação
GRUPOS
0
10
20
30
40
50

7) Discussão

A ausência de estudos epidemiológicos das manifestações orais e dentárias dos pacientes com IDP dificulta muito o entendimento acerca da realidade da freqüência destes achados. Nosso estudo, na tentativa de definir a freqüência das manifestações orais e dentárias nas IDPs com alterações de número e/ou função de células fagocitárias, desejava buscar uma resposta para este questionamento, além de impulsionar uma discussão sobre a necessidade de uma abordagem multidisciplinar e integral destes pacientes, que podem ser encontrados na prática odontológica.

Através da análise destes 632 artigos, pudemos perceber o quanto a cavidade oral é negligenciada durante o exame clínico dos pacientes, em estudos conduzidos fora do contexto da pesquisa odontológica. Uma vez que a grande maioria dos artigos é escrita pelas equipes médicas diretamente envolvidas nos casos, a falta de treinamento e até mesmo o esquecimento do exame da cavidade oral comprometeram diretamente os nossos resultados. Em algumas doenças, como na Síndrome de Papillon-Lefrévre, por exemplo, na qual a doença periodontal faz parte da tríade diagnóstica, observamos que em 8% dos pacientes analisados não houve relato de doença periodontal.

Esta carência é tanto mais grave quanto, entre os muitos sítios que mais freqüentemente podem ser afetados pelas IDPs, a cavidade oral, como a pele, são especialmente ricos em informações, além de serem acessíveis ao profissional treinado, sem risco nem desconforto para o paciente, através de métodos não-invasivos.

Já que as manifestações orais foram encontradas em 37,9% dos pacientes analisados, entendemos que, o cirurgião-dentista, sobretudo o

periodontista e o estomatologista, deve ser visto como um elemento importante na abordagem do exame da cavidade oral, pois o diagnóstico precoce de uma infecção intra-oral ou até mesmo de uma anomalia dentária, pode auxiliar no diagnóstico e no tratamento, com o intuito de prevenir ou minimizar os agravos.

Além disso, de acordo com os resultados encontrados neste presente estudo, de acordo com a freqüência de doença periodontal relatada, fica claro que a doença periodontal pode ser entendida como um sinal de alerta de IDP por deficiência de células fagocitárias.

A gravidade de alguns casos clínicos analisados pode ter interferido nos resultados encontrados, visto que como algumas doenças cursam com óbito ainda uma idade bastante precoce, a abordagem das infecções de cavidade oral destas crianças acaba sendo pouco valorizada dada a gravidade do quadro clínico das demais infecções sistêmicas.

Além disso, muitos dos relatos de casos avaliados na busca realizada foram descritos antes mesmo do início da dentição decídua, impossibilitando, portanto, o envolvimento da saúde do tecido periodontal, geralmente associado às doenças de fagócitos.

Outro aspecto a ressaltar é a freqüência das diferentes manifestações orais e dentárias encontrados na nossa análise. Alguns dados são considerados sintomas ou evolução do quadro de uma doença e foram agrupados separadamente, para evitar a supervalorização de uma determinada manifestação em detrimento de outra. Por exemplo, a perda precoce de dentes decíduos, a perda de osso alveolar, a mobilidade dentária são evoluções clínicas esperadas da doença periodontal e estes pacientes com estas manifestações, provavelmente apresentavam um quadro de periodontite.

Outras manifestações como envolvimento da saúde oral e estomatite, considerados nos resultados por aparecerem relatados com esta terminologia nos artigos, não trouxeram nenhuma conclusão útil, já que se tratam de termos abrangentes e inespecíficos. A estomatite, por exemplo, pode ser por espécies de *Candida*, por herpes ou simplesmente um quadro de estomatite aftosa recorrente. Cabe lembrar ainda que nos artigos onde estivesse relatada estomatite aftosa recorrente, a mesma foi agrupada junto com as aftas.

Nossos resultados foram, em geral, condizentes com os descritos na literatura até o presente momento (Szczawinska-Poplonik *et al*, 2009; Aktinson *et al* 2000).

De acordo com Szczawinska-Poplonik e colaboradores (2009), as manifestações orais e dentárias comumente descritas na Síndrome de Kostmann incluem a gengivite, a periodontite e a perda de osso alveolar. Nossos resultados também destacam a doença periodontal e a gengivite, porém encontramos um número significativo de pacientes com úlceras aftosas (61%). Já nas neutropenias cíclicas, as úlceras orais encontradas em 41% dos pacientes também são achados característicos em outros estudos (Szczawinska-Poplonik *et al*, 2009; Aktinson *et al*, 2000)

A doença periodontal e a gengivite foram relatadas em quantidade expressiva nas deficiências de adesão leucocitária (LAD-1, LAD-2 e LAD-3), nas neutropenias congênita e cíclica e na Síndrome de Papillon-Lefrève, corroborando, portanto, com as descrições de Szczawinska-Poplonik e colaboradores (2009) e de Aktinson e colaboradores (2000).

A presença de candidíase oral nos pacientes com Síndrome de Hiper IgE relatada por Szczawinska-Poplonik e colaboradores (2009) também foi

encontrada em 27,4% dos casos de Síndrome de Hiper IgE autossômica dominante. Além da candidíase, a retenção prolongada de dentes decíduos, característica marcante da síndrome e presente no *score* para Hiper IgE proposta por Freeman e colaboradores (2008), também foi um dado bastante expressivo na população estudada, correspondendo a 67,9% dos casos analisados.

As úlceras orais, destacadas por Szczawinska-Poplonik e colaboradores (2009) e por Aktinson e colaboradores (2000) como manifestações comumente encontradas na DGC e nas neutropenias. No nosso estudo, as úlceras orais, além de freqüentes nas neutropenias e na DGC ligada ao X também foram relatadas nas deficiências de adesão leucocitária do tipo 1.

Nosso trabalho veio, acima de tudo, tornar mais claro que alterações orais e dentárias não são igualmente freqüentes nas diferentes formas de IDP, nem mesmo quando se toma uma categoria específica de IDP, como o grupo V da classificação mais recente. O exame da cavidade oral será especialmente útil no caso de alguns pacientes, portadores das patologias específicas listadas acima. Nas demais, seja pela sua raridade, seja pela raridade das alterações acessíveis ao odontólogo, a sua busca terá provavelmente pouca utilidade.

Uma conseqüência mais geral do trabalho de revisão sistemática realizado por nós é que a evolução rápida dos estudos moleculares afetou de muitas maneiras a descrição dos pacientes com IDP. Na literatura mais recente, a ênfase é na caracterização de mutações específicas, muito mais do que na caracterização de sinais e sintomas clínicos. Com isso, muitas referências simplesmente escapam à detecção, já que os termos de busca

eficazes são os que identificam o gene mutado, e não os que identificam a doença definida em termos clínicos, anatomo-clínicos ou epidemiológicos.

Tomamos o cuidado de confirmar, sistematicamente, que o procedimento de busca mais específico adotado por nós, com base na pesquisa de literatura sobre mutações, recuperava também a literatura que tinha sido localizada através da abordagem convencional, com termos de busca abrangentes. Embora seja este o caso, o número de publicações que foram recuperadas na busca específica e que escaparam da busca abrangente pode ser a grande maioria das referências compiladas na nossa análise final. Isto deve servir de alerta para os que empreenderem estudos semelhantes, já que reflete as limitações dos sistemas eletrônicos de busca, assim como a rápida evolução da pesquisa biomédica.

A rápida evolução da medicina, resultante de estudos moleculares, também nos impede de utilizar com a mesma eficiência informação publicada em décadas anteriores a 1980. Isto se explica porque muitas das IDPs hoje reconhecidas foram descritas de 1980 para cá, mas também porque na fase inicial a caracterização de cada IDP era predominantemente clínica, e, não havendo identificação de mutações ou de defeitos moleculares bem-caracterizados, as referências correspondentes simplesmente escapam à detecção, sempre que a busca for feita em termos de mutações específicas. Isto não seria problema se todo trabalho atual com métodos moleculares incluísse no título ou resumo os nomes já reconhecidos das doenças, o que permitiria recuperá-los desta forma. Isto, no entanto, nem sempre ocorre, nem evita os problemas resultantes de mudanças inevitáveis nos nomes das

doenças, através dos avanços do conhecimento que são consolidados em classificações revistas periodicamente.

Com estas ressalvas em mente, consideramos que o presente estudo representa um primeiro esforço de disponibilizar para o profissional de Saúde no Brasil, e especialmente para aquele que se envolve no atendimento a pacientes pediátricos, seja em Odontologia, seja em diferentes especialidades da Medicina Clínica, um retrato quantitativo e preciso da literatura científica existente sobre alterações da cavidade oral em IDPs com deficiências de fagócitos.

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ANEXOS

TABLE V. Congenital defects of phagocyte number, function, or both

Disease	Affected cells	Affected function	Associated features	Inheritance	Gene defect—presumed pathogenesis	Relative frequency among PIDs
1.-2. Severe congenital neutropenias	N	Myeloid differentiation	Subgroup with myelodysplasia	AD	*ELA2*: mistrafficking of elastase	Rare
	N	Myeloid differentiation	B/T lymphopenia	AD	*GFI1*: repression of elastase	Extremely rare
3. Kostmann disease	N	Myeloid differentiation	Cognitive and neurological defects*	AR	*HAX1*: control of apoptosis	Rare
4 Neutropenia with cardiac and urogenital malformations	N + F	Myeloid differentiation	Structural heart defects, urogenital abnormalities, and venous angiectasias of trunks and limbs	AR	*G6PC3*: abolished enzymatic activity of glucose-6-phosphatase and enhanced apoptosis of N and F	Very rare
5 Glycogen storage disease type 1b	N + M	Killing, chemotaxis, O_2^- production	Fasting hypoglycemia, lactic acidosis, hyperlipidemia, hepatomegaly, neutropenia	AR	*G6PT1*: Glucose-6-phosphate transporter 1	Very rare
6. Cyclic neutropenia	N	?	Oscillations of other leukocytes and platelets	AD	*ELA2*: mistrafficking of elastase	Very rare
7. X-linked neutropenia/ myelodysplasia	N + M	?	Monocytopenia	XL	*WAS*: Regulator of actin cytoskeleton (loss of autoinhibition)	Extremely rare
8. P14 deficiency	N+L Mel	Endosome biogenesis	Neutropenia Hypogammaglobulinemia ↓CD8 cytotoxicity Partial albinism Growth failure	AR	*MAPBPIP*: Endosomal adaptor protein 14	Extremely rare
9. Leukocyte adhesion deficiency type 1	N + M + L + NK	Adherence Chemotaxis Endocytosis T/NK cytotoxicity	Delayed cord separation, skin ulcers Periodontitis Leukocytosis	AR	*ITGB2*: Adhesion protein	Very rare
10. Leukocyte adhesion deficiency type 2	N + M	Rolling chemotaxis	Mild LAD type 1 features plus hh-blood group plus mental and growth retardation	AR	*FUCT1*: GDP-Fucose transporter	Extremely rare
11. Leukocyte adhesion deficiency type 3	N + M + L + NK	Adherence	LAD type 1 plus bleeding tendency	AR	*KINDLIN3*: Rap1-activation of β1-3 integrins	Extremely rare
12. Rac 2 deficiency	N	Adherence Chemotaxis O_2^- production	Poor wound healing, leukocytosis	AD	*RAC2*: Regulation of actin cytoskeleton	Extremely rare: Regulation of actin cytoskeleton
13. β-Actin deficiency	N + M	Motility	Mental retardation, short stature	AD	*ACTB*: Cytoplasmic actin	Extremely rare
14. Localized juvenile periodontitis	N	Formylpeptide-induced chemotaxis	Periodontitis only	AR	*FPR1*: Chemokine receptor	Very rare
15. Papillon-Lefèvre syndrome	N + M	Chemotaxis	Periodontitis, palmoplantar hyperkeratosis†	AR	*CTSC*: Cathepsin C activation of serine proteases	Very rare
16. Specific granule deficiency	N	Chemotaxis	N with bilobed nuclei	AR	*CEBPE*: myeloid transcription factor	Extremely rare
17. Shwachman-Diamond syndrome	N	Chemotaxis	Pancytopenia, exocrine pancreatic insufficiency, chondrodysplasia	AR	*SBDS*	Rare
18. X-linked chronic granulomatous disease (CGD)	N + M	Killing (faulty O_2^- production)	McLeod phenotype in a subgroup of patients	XL	*CYBB*: Electron transport protein (gp91phox)	Relatively common

(Continued)

TABLE V. *(Continued)*

sease	Affected cells	Affected function	Associated features	Inheritance	Gene defect—presumed pathogenesis	Relative frequency among PIDs
9.- 21. Autosomal CGDs	N + M	Killing (faulty O_2^- production)		AR	*CYBA*: Electron transport protein (p22phox) *NCF1*: Adapter protein (p47phox) *NCF2*: Activating protein (p67phox)	Relatively common
2. IL-12 and IL-23 receptor β1 chain deficiency	L + NK	IFN-γ secretion	Susceptibility to mycobacteria and *Salmonella*	AR	*IL12RB1*: IL-12 and IL-23 receptor β1 chain	Rare
3. IL-12p40 deficiency	M	IFN-γ secretion	Susceptibility to mycobacteria and *Salmonella*	AR	*IL12B*: subunit of IL12/IL23	Very rare
4. IFN-γ receptor 1 deficiency	M + L	IFN-γ binding and signaling	Susceptibility to mycobacteria and *Salmonella*	AR, AD	*IFNGR1*: IFN-γR ligand binding chain	Rare
5. IFN-γ receptor 2 deficiency	M + L	IFN-γ signaling	Susceptibility to mycobacteria and *Salmonella*	AR	*IFNGR2*: IFN-γR accessory chain	Very rare
6. STAT1 deficiency (2 forms)	M + L	IFN α/β, IFN-γ, IFN-λ, and IL-27 signaling	Susceptibility to mycobacteria, *Salmonella* and *viruses*	AR	*STAT1*	Extremely rare
7. AD hyper-IgE	L+M+N+ epithelial	IFN-γ signaling	Susceptibility to mycobacteria and *Salmonella*	AD	*STAT1*	Extremely rare
8. AR hyper-IgE (TYK2 deficiency)	L+M+N+ others	IL-6/10/22/23 signaling IL-6/10/12/ 23/IFN-α/ IFN-β signaling	Distinctive facial features (broad nasal bridge); eczema; osteoporosis and fractures; scoliosis; failure/delay of shedding primary teeth; hyperextensible joints; bacterial infections (skin and pulmonary abscesses/ pneumatoceles) caused by *Staphylococcus aureus*; candidiasis Susceptibility to intracellular bacteria (mycobacteria, *Salmonella*), *Staphylococcus*, and viruses.	AD AD	*STAT3* *TYK2*	Rare Extremely rare
9. Pulmonary alveolar proteinosis	Alveolar macrophages	GM-CSF signaling	Alveolar proteinosis	biallelic mutations in pseudoautosomal gene	*CSF2RA*	extremely rare

CTB, Actin beta; *AD*, autosomal-dominant; *AR*, autosomal-recessive inheritance; *CEBPE*, CCAAT/Enhancer-binding protein epsilon; *CTSC*, cathepsin C; *CYBA*, cytochrome b pha subunit; *CYBB*, cytochrome b beta subunit; *ELA2*, elastase 2; *IFN*, interferon; *IFNGR1*, interferon-gamma receptor subunit 1; *IFNGR2*, interferon-gamma receptor subunit 2; *12B*, interleukin-12 beta subunit; *IL12RB1*, interleukin-12 receptor beta 1; *F*, fibroblasts; *FPR1*, formylpeptide receptor 1; *FUCT1*, fucose transporter 1; *GFI1*, growth factor dependent 1; *HAX1*, HLCS1-associated protein X1; *ITGB2*, integrin beta-2; *L*, lymphocytes; *M*, monocytes-macrophages; *MAPBPIP*, MAPBP-interacting protein; *Mel*, elanocytes; *N*, neutrophils; *NCF1*, neutrophil cytosolic factor 1; *NCF2*, neutrophil cytosolic factor 2; *NK*, natural killer cells; *SBDS*, Shwachman-Bodian-Diamond syndrome; *AT*, signal transducer and activator of transcription; *XL*, X-linked inheritance.

Cognitive and neurologic defects are observed in a fraction of patients.

Periodontitis may be isolated.

APÊNDICES

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1. Severe congenital neutropenia resulting from G6PC3 deficiency with increased neutrophil CXCR4 expression and myelokathexis.
McDermott DH, De Ravin SS, Jun HS, Liu Q, Priel DA, Noel P, Takemoto CM, Ojode T, Paul SM, Dunsmore KP, Hilligoss D, Marquesen M, Ulrick J, Kuhns DB, Chou JY, Malech HL, Murphy PM.
Blood. 2010 Oct 14;116(15):2793-802. Epub 2010 Jul 8.
PMID: 20616219 [PubMed - indexed for MEDLINE]

2. A novel G6PC3 homozygous 1-bp deletion as a cause of severe congenital neutropenia.
Aróstegui JI, de Toledo JS, Pascal M, García C, Yagüe J, Díaz de Heredia C.
Blood. 2009 Aug 20;114(8):1718-9. No abstract available.
PMID: 19696212 [PubMed - indexed for MEDLINE] **Free Article**

3. Acute lymphoblastic leukemia following severe congenital neutropenia or de novo ALL?
Valera ET, Brassesco MS, Germeshausen M, Silveira Vda S, Queiroz RG, Roxo P, Scrideli CA, de Menezes UP, Ferriani V, Tone LG.
Leuk Res. 2009 Sep;33(9):e139-42. Epub 2009 Apr 26.
PMID: 19398129 [PubMed - indexed for MEDLINE]

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Search: Neutropenia with cardiac and urogenital malformations

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Results: 3

1. A syndrome with congenital neutropenia and mutations in G6PC3.
Boztug K, Appaswamy G, Ashikov A, Schäffer AA, Salzer U, Diestelhorst J, Germeshausen M, Brandes G, Lee-Gossler J, Noyan F, Gatzke AK, Minkov M, Greil J, Kratz C, Petropoulou T, Pellier I, Bellanné-Chantelot C, Rezaei N, Mönkemöller K, Irani-Hakimeh N, Bakker H, Gerardy-Schahn R, Zeidler C, Grimbacher B, Welte K, Klein C.
N Engl J Med. 2009 Jan 1;360(1):32-43.
PMID: 19118303 [PubMed - indexed for MEDLINE] **Free PMC Article**

2. The many causes of severe congenital neutropenia.
Dale DC, Link DC.
N Engl J Med. 2009 Jan 1;360(1):3-5. No abstract available.
PMID: 19118300 [PubMed - indexed for MEDLINE]

3. Toxicity of high-dose ifosfamide in children.
Davies SM, Pearson AD, Craft AW.
Cancer Chemother Pharmacol. 1989;24 Suppl 1:S8-10.
PMID: 2503259 [PubMed - indexed for MEDLINE]

PubMed

Search: Glucose-6- phosphate transporter 1 or G6PT1 AND case reports

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SLC37A4 (G6PT1) solute carrier family 37 (glucose-6-phosphate transporter), member 4 [Homo sapiens]

g6pt1 in Homo sapiens | Mus musculus | Rattus norvegicus | All 6 Gene records

Results: 6

1. A patient with common glycogen storage disease type Ib mutations without neutropenia or neutrophil dysfunction.
Martens DH, Kuijpers TW, Maianski NA, Rake JP, Smit GP, Visser G.
J Inherit Metab Dis. 2006 Feb;29(1):224-5.
PMID: 16601899 [PubMed - indexed for MEDLINE]

2. Glycogen storage disease type Ib without neutropenia generated by a novel splice-site mutation in the glucose-6-phosphate translocase gene.
Angaroni CJ, Labrune P, Petit F, Sastre D, Capra AE, Dodelson de Kremer R, Argaraña CE.
Mol Genet Metab. 2006 May;88(1):96-9. Epub 2006 Feb 21.
PMID: 16490377 [PubMed - indexed for MEDLINE]

3. Intestinal glucose transport: evidence for a membrane traffic-based pathway in humans.
Santer R, Hillebrand G, Steinmann B, Schaub J.
Gastroenterology. 2003 Jan;124(1):34-9.
PMID: 12512027 [PubMed - indexed for MEDLINE]

4. Novel missense mutation (Y24H) in the G6PT1 gene causing glycogen storage disease type 1b.
Yuen YP, Cheng WF, Tong SF, Chan YT, Chan YW, Lam CW.
Mol Genet Metab. 2002 Nov;77(3):249-51.
PMID: 12409273 [PubMed - indexed for MEDLINE]

5. Multiple transport protein defects in a patient with glycogen storage disease type 1: GSD 1b/1c beta.
Hawkins RA, Kamath KR, Scott HM, Burchell A.
J Inherit Metab Dis. 1995;18(5):558-66.
PMID: 8598636 [PubMed - indexed for MEDLINE]

6. Glycogen storage disease type 1b due to a defect of glucose-6-phosphate translocase.
Narisawa K, Otomo H, Igarashi Y, Arai N, Otake M, Tada K, Kuzuya T.
J Inherit Metab Dis. 1982;5(4):227-8.
PMID: 6133035 [PubMed - indexed for MEDLINE]

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Search: Glucose-6- phosphate transporter 1 or G6PT1 AND case reports

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See 14 articles about **SLC37A4 (G6PT1)** gene function

SLC37A4 (G6PT1) solute carrier family 37 (glucose-6-phosphate transporter), member 4 [Homo sapiens]

g6pt1 in Homo sapiens | Mus musculus | Rattus norvegicus | All 6 Gene records

Results: 6

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J Inherit Metab Dis. 1982;5(4):227-8.
PMID: 6133035 [PubMed - indexed for MEDLINE]

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Search: Glucose-6- phosphate transporter 1 or G6PT1

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Results: 51

1. Carbohydrate-response-element-binding protein (ChREBP) and not the liver X receptor α (LXRα) mediates elevated hepatic lipogenic gene expression in a mouse model of glycogen storage disease type 1.
Grefhorst A, Schreurs M, Oosterveer MH, Cortés VA, Havinga R, Herling AW, Reijngoud DJ, Groen AK, Kuipers F.
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2. Altered redox state of luminal pyridine nucleotides facilitates the sensitivity towards oxidative injury and leads to endoplasmic reticulum stress dependent autophagy in HepG2 cells.
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PMID: 18267120 [PubMed - indexed for MEDLINE]

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Walker EA, Ahmed A, Lavery GG, Tomlinson JW, Kim SY, Cooper MS, Ride JP, Hughes BA, Shackleton CH, McKiernan P, Elias E, Chou JY, Stewart PM.
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10. Necrosis induction in glioblastoma cells reveals a new "bioswitch" function for the MT1-MMP/G6PT signaling axis in proMMP-2 activation versus cell death decision.
Belkaid A, Fortier S, Cao J, Annabi B.
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A patient with common glycogen storage disease type Ib mutations without neutropenia or neutrophil dysfunction.

13. Martens DH, Kuijpers TW, Maianski NA, Rake JP, Smit GP, Visser G.
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14. Angaroni CJ, Labrune P, Petit F, Sastre D, Capra AE, Dodelson de Kremer R, Argaraña CE.
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J Inherit Metab Dis. 1982;5(4):227-8.
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Search: Cyclic neutropenia syndrome and case reports

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Filter your results: All (20)
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Results: 20

1. Image of the month. Albinism, cyclic neutropenia, and ceroid pigment in the liver.
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19. [Cyclic neutropenia. Detection of a mutation in the gene for neutrophil elastase (ELA2)].
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Search: Cyclic neutropenia and case reports and ELA2

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ELANE-Related Neutropenia Source: GeneReviews

ELANE-related neutropenia includes congenital neutropenia and cyclic neutropenia, both of which are primary hematologic disorders characterized by recurrent fever, skin and oropharyngeal inflammation (i.e., mouth ulcers, gingivitis, sinusitis, and pharyngitis), and cervical adenopathy.

Genetic Counseling Management

Results: 2

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Genetic Counseling Management

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Genetic Counseling Management

Results: 16

1. The Wiskott-Aldrich syndrome: The actin cytoskeleton and immune cell function.
Blundell MP, Worth A, Bouma G, Thrasher AJ.
Dis Markers. 2010;29(3-4):157-75. Review.
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2. Activating WASP mutations associated with X-linked neutropenia result in enhanced actin polymerization, altered cytoskeletal responses, and genomic instability in lymphocytes.
Westerberg LS, Meelu P, Baptista M, Eston MA, Adamovich DA, Cotta-de-Almeida V, Seed B, Rosen MK, Vandenberghe P, Thrasher AJ, Klein C, Alt FW, Snapper SB.
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3. WASP: a key immunological multitasker.
Thrasher AJ, Burns SO.
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4. New insights into the biology of Wiskott-Aldrich syndrome (WAS).
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Biol Blood Marrow Transplant. 2009 Jan;15(1 Suppl):84-90. Review.
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PMID: 17724125 [PubMed - indexed for MEDLINE] **Free PMC Article**

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Constitutively activating mutation in WASP causes X-linked severe congenital neutropenia.
16. Devriendt K, Kim AS, Mathijs G, Frints SG, Schwartz M, Van Den Oord JJ, Verhoef GE, Boogaerts MA, Fryns JP, You D, Rosen MK, Vandenberghe P.
Nat Genet. 2001 Mar;27(3):313-7.
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Role of neutrophil elastase in bone marrow failure syndromes: molecular genetic revival of the chalone hypothesis.
15. Horwitz M, Benson KF, Duan Z, Person RE, Wechsler J, Williams K, Albani D, Li FQ.
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PMID: 12483111 [PubMed - indexed for MEDLINE]

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Nat Genet. 2001 Mar;27(3):313-7.
PMID: 11242115 [PubMed - indexed for MEDLINE]

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Results: 3

1. The Wiskott-Aldrich syndrome: The actin cytoskeleton and immune cell function.
Blundell MP, Worth A, Bouma G, Thrasher AJ.
Dis Markers. 2010;29(3-4):157-75. Review.
PMID: 21178275 [PubMed - indexed for MEDLINE]

2. Two novel activating mutations in the Wiskott-Aldrich syndrome protein result in congenital neutropenia.
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Blood. 2006 Oct 1;108(7):2182-9. Epub 2006 Jun 27.
PMID: 16804117 [PubMed - indexed for MEDLINE] **Free Article**

3. Role of neutrophil elastase in bone marrow failure syndromes: molecular genetic revival of the chalone hypothesis.
Horwitz M, Benson KF, Duan Z, Person RE, Wechsler J, Williams K, Albani D, Li FQ.
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Results: 112

1. The Wiskott-Aldrich syndrome: The actin cytoskeleton and immune cell function.
Blundell MP, Worth A, Bouma G, Thrasher AJ.
Dis Markers. 2010;29(3-4):157-75. Review.
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Refinement of the PARK3 locus on chromosome 2p13 and the analysis of 14 candidate genes.

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PMID: 7628435 [PubMed - indexed for MEDLINE] **Free PMC Article**

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1. Pediatr Blood Cancer. 2008 Sep;51(3):436-8.

Lethal graft-versus-host disease in congenital neutropenia caused by p14 deficiency after allogeneic bone marrow transplantation from an HLA-identical sibling.

Bohn G, Hardtke-Wolenski M, Zeidler C, Maecker B, Sauer M, Sykora KW, Grigull L, Welte K, Klein C.

Department of Pediatric Hematology and Oncology, Hannover Medical School, OE 6780, Carl-Neuberg-Str. 1, 30625 Hannover, Germany.

Abstract
The molecular heterogeneity of severe congenital neutropenia (SCN) is increasingly recognized and may influence the risk-benefit assessment of therapeutic strategies. We report on a patient with p14 deficiency who succumbed to severe grade IV graft-versus-host disease (GvHD) after a human leukocyte antigen-identical bone marrow transplantion (BMT) from a sibling donor. Before BMT, in vitro generated p14-deficient dendritic cells showed a markedly elevated tumor necrosis factor (TNF-) alpha production upon toll-like receptor stimulation. We hypothesize that p14 deficiency predisposes to GvHD through increased TNF-alpha production. Adequate genetic testing is needed to prospectively assess potential risk factors for GvHD in defined SCN subgroups.

PMID: 18523989 [PubMed - indexed for MEDLINE]

Publication Types, MeSH Terms, Substances

2. Prenat Diagn. 2005 Dec;25(12):1133-7.

Prenatal diagnosis of monosomy 4p14-->pter and trisomy 11q25-->qter: clinical presentations and outcomes.

Peng HH, Wang TH, Chao AS, Chang YL, Chang SD, Soong YK.

Department of Obstetrics and Gynecology, Chang Gung Memorial Hospital, Lin-ko Medical Center, Kwei-Shan, Tao-Yuan, Taiwan.

Abstract
We present the case of a pregnant woman with low free beta-HCG in maternal serum Down syndrome screening that led to prenatal diagnosis of a fetus with 46,XY,der(4)t(4;11)(p14; q25). This chromosomal aneuploidy resulted from unbalanced segregation of a paternal balanced translocation, t(4;11)(p14;q25). Prenatal ultrasound revealed intrauterine growth restriction, cleft lip and palate, a thick nuchal fold, a single umbilical artery, and pyelectasis. Array-based comparative genomic hybridization and short tandem repeat markers further located the exact breakpoint of translocation. The woman had her pregnancy terminated at 23 weeks of gestational age. The proband had general appearance of Wolf-Hirschhorn syndrome and some unique findings, including single umbilical artery, severe immunoglobulin deficiency, scalp defect, and underlying bony defect. Our case underscores the importance of fetal karyotyping when low maternal serum free beta-HCG is found. It also adds information on the fetal presentations of monosomy 4p14-->pter and trisomy 11q25-->qter.

PMID: 16231301 [PubMed - indexed for MEDLINE]

Publication Types, MeSH Terms, Substances

3. Eur J Hum Genet. 2005 Apr;13(4):409-13.

Combination of WAGR and Potocki-Shaffer contiguous

deletion syndromes in a patient with an 11p11.2-p14 deletion.

Brémond-Gignac D, Crolla JA, Copin H, Guichet A, Bonneau D, Taine L, Lacombe D, Baumann C, Benzacken B, Verloes A.

Department of Ophthalmology, Robert Debré Hospital, AP-HP, Paris, France.

Abstract

Aniridia, Wilms tumor, genitourinary abnormalities, growth and mental retardation are the cardinal features of the WAGR 11p13 deletion syndrome. The Potocki-Schaffer syndrome or proximal 11p deletion syndrome (previously DEFECT11 syndrome) is a contiguous gene syndrome associated with deletions in 11p11.2, principal features of which are multiple exostoses and enlarged parietal foramina. Mental handicap, facial dysmorphism and craniosynostosis may also be associated. We report a patient with combined WAGR and Potocki-Shaffer syndromes, and obesity. She presented with aniridia, cataract, nystagmus, corneal ulcers and bilateral congenital ptosis. A left nephroblastoma was detected at 15 months. Other features included moderate developmental delay, growth deficiency, facial dysmorphism, multiple exostoses and cranial lacunae. High-resolution and molecular cytogenetics confirmed a del(11)(p11.2p14.1) deletion with a proximal breakpoint between the cosmid DO8153 and the BAC RP11-104M24 to a distal breakpoint between cosmids CO8160 (D11S151) and F1238 (D11S1446). The deletion therefore includes EXT2, ALX4, WT1 and PAX6. This case appears to be the second patient reported with this combined deletion syndrome and confirms the association of obesity in the WAGR spectrum, a feature previously reported in four cases, and for which the acronym WAGRO has been suggested. Molecular and follow-up data on the original WAGRO case are briefly presented.

PMID: 15702131 [PubMed - indexed for MEDLINE] **Free Article**

Publication Types, MeSH Terms

4. Am J Med Genet. 2002 Jul 15;110(4):353-8.

Unbalanced translocation (3;5)(q26.1;p14): a clinical report.

Rossi M, Di Micco P, Perone L, De Brasi D, Guzzetta V, Andreucci MV, Vega GR, Marzano MG, Iaccarino E, Andria G.

Department of Pediatrics, Federico II University, Naples, Italy.

Abstract

A patient with a multiple congenital anomalies/mental retardation (MCA/MR) syndrome had an unbalanced translocation (3;5)(q26.1;p14), causing partial 5p monosomy and partial 3q trisomy. The phenotype observed in this patient results from the combination of those described in the isolated dup(3q) and del(5p) syndromes. Some clinical features of this patient are shared by the Smith-Lemli-Opitz syndrome (SLOS), a well-known MCA/MR syndrome due to the deficiency of 7-dehydrocholesterol reductase (DHCR7). We review the previously reported cases of chromosomal anomalies with clinical features suggesting SLOS.

PMID: 12116209 [PubMed - indexed for MEDLINE]

Publication Types, MeSH Terms

5. Hum Genet. 2000 Nov;107(5):415-32.

Interchromosomal insertions. Identification of five cases and a review.

Van Hemel JO, Eussen HJ.

Department of Clinical Genetics, University Hospital Dijkzigt, Erasmus University, Rotterdam, The Netherlands. vanhemel@kgen.fgg.eur.nl

Abstract

In five families with questionable chromosome rearrangements, we identified an interchromosomal insertion by fluorescent in situ hybridization (FISH). In case 1 with a dir ins (5;11)(p14;q14q24) in three generations, the mentally retarded and microcephalic proband showed a 5p14-->pter deletion. In case 2, a duplication (13)(q21.31--> q31.2) combined with a deletion (11)(q14-->q22) segregated from a reciprocal ins(11;13)(q14q122)(q21.32q31.2), causing a mixed phenotype with psychomotor retardation, caput quadratum, choanal atresia, and pes equinovarus. In case 3, a dir ins (18;5)(q21.3;p13.1p14) was associated with spontaneous abortions, in case 4, the proband with mental retardation, microcephaly, and a heart defect showed a pure trisomy of (12)(q13-->q15), which had segregated from a carrier of an ins (18;12)(p11.3;q13q15). In case 5, a duplication of (10)(q26.3-->q25.2) segregated from an inv

ins(5;10)(q15;q26.3q25.2), which was passed on directly from a mother to her son,with mental retardation. In all families the elucidation of the insertional translocation (IT) considerably increased the associated genetic risks of carriers. For the review, we collected data from 81 articles on 87 IT probands on ascertainment, origin, familial transmittance, progeny, and genetic risks of IT carriers. We also discussed the recombinant chromosomes and complex rearrangements associated with ITs, and listed chromosome regions occurring solely as deletions, or solely as duplications, or as both to facilitate genotype/phenotype correlations. We conclude that ITs are rare chromosomal rearrangements with an 1:80,000 incidence, of which nearly 80% were referred because of congenital abnormalities and mental retardation. A maternal origin was seen in 59.5%, a paternal origin in 26.6%, and 13.9% were de novo. No notable difference in fertility between male and female IT carriers was noticed. Bias of ascertainment was excluded in 15 familial cases and led to an estimate of the genetic risks for IT carriers of 32.0-36.0%. The mean size of the inserted regions occurring solely as duplications (n=39) measures 0.96% of the haploid autosomal length (HAL), and of regions solely occurring as deletions (n=14) 0.47% HAL. In the families where both aneusomies occurred, the size of the insertions ranged between 0.22 and 1.21% HAL. Overall, the findings fit with the general idea that a surplus of genetic material is tolerated more easily than a deficiency.

PMID: 11140939 [PubMed - indexed for MEDLINE]

Publication Types, MeSH Terms, Substances

6. Am J Med Genet. 1998 May 26;77(4):257-60.

Partial trisomy 1q with growth hormone deficiency and normal intelligence.

Schorry EK, Dietrich KN, Saal HM, Blough RI, Dey S, Chernausek S, Milatovich-Cherry A.

Division of Human Genetics, Children's Hospital Medical Center, Cincinnati, Ohio 45229, USA.

Abstract

We present two sibs with partial trisomy 1 (q31.1-q32.1) due to a familial insertion. Patient 1 is a girl who presented at age 9 months with minor anomalies, short stature, and normal psychomotor development. Karyotype was 46,XX,der(4)ins(4;1) (p14;q31.1q32.1)pat. The father had a balanced inverted insertion of 1q into 4p, with karyotype 46,XY,ins(4;1)(p14;q31.1q32.1). At age 5 years, patient 1 was found to have short stature with documented growth hormone deficiency and ectopic pituitary. Her growth velocity responded well to treatment with growth hormone. Cognitive testing at 5 9/12 years showed normal intelligence with an IQ of 90. Patient 2, the brother of patient 1, presented with intrauterine growth retardation. He has the same chromosomal insertion as his sister, with partial trisomy 1q. We suggest that there is a recognizable phenotype of trisomy 1(q31.1-q32.1) which includes prenatal and postnatal growth retardation, narrow palpebral fissures, microphthalmia, microstomia, pituitary abnormalities, and normal intelligence in some individuals.

PMID: 9600731 [PubMed - indexed for MEDLINE]

Publication Types, MeSH Terms, Substances

7. Electroencephalogr Clin Neurophysiol. 1992 Sep-Oct;84(5):433-9.

Central sensory and motor conduction in vitamin B12 deficiency.

Di Lazzaro V, Restuccia D, Fogli D, Nardone R, Mazza S, Tonali P.

Department of Neurology, Catholic University, Rome, Italy.

Abstract

Four patients with subacute combined degeneration were studied through upper and lower limb SEPs recorded with a non-cephalic reference montage and through cortical and spinal magnetic stimulation. Clinical signs were confined to the lower limbs in 3 patients; the remaining patient presented only paraesthesiae in 4 limbs. Median nerve SEPs showed a normal cervical N13 response with a significant increase of central conduction time concerning exclusively the P9-P14 interpeak interval. Central motor conduction to upper and lower limb muscles was abnormal. Nerve conduction studies provided no evidence of peripheral nerve involvement. These electrophysiological findings suggest that in vitamin B12 deficiency the higher segments of the cervical cord are usually affected first and that central sensory and motor conduction studies are sensitive methods for detecting such damage.

PMID: 1382952 [PubMed - indexed for MEDLINE]

Publication Types, MeSH Terms

Rev Neurol (Paris). 1991;147(11):723-6.

[Course of central somatosensory conduction in a case of vitamin B12 deficiency].

[Article in French]

Delberghe X, Brunko E, Zegers de Beyl D.

Service de Neurologie, Hôpital Erasme, Bruxelles, Belgique.

Abstract

We report clinical and neurophysiological data of a 58 year-old man with vitamin B12 deficiency and a 6 years follow-up. The initial clinical disorders did not permit a clear distinction between peripheral and central nervous system disease. Detailed analysis of the somatosensory central conduction time (measured from onset latencies of N11 and N20) showed clear evidence of slowed down conduction at spinal cervical level (reflected by the N11-P14 interval) whereas supraspinal conduction (as shown by the P14-N20 interval) was normal. The patient's condition improved under treatment, and the somatosensory central conduction time was progressively shortened. After 6 years of treatment, the neurological examination and the somatosensory central conduction interval were normal. However, analysis of the spinal conduction still showed slowed down conduction.

PMID: 1775826 [PubMed - indexed for MEDLINE]

Publication Types, MeSH Terms

9. J Genet Hum. 1983 Mar;31(1):31-6.

[Chromosome 11 and cancer].

[Article in French]

Gregoire MJ, Pernot C, Himont F, Pierson M, Gilgenkrantz S.

Abstract

Two cases with chromosome 11 anomaly related to cancer are reported. The first one has a pericentric inversion (inv. p14 q12) with sympathoblastoma and Ondine's curse. The second one has a deletion (11p13) with aniridia and catalase deficiency but without Wilms tumor at two year of age. Retinoblastoma, nephroblastoma and sympathoblastoma may be related to genome modification. The mechanism of oncogenesis are discussed.

PMID: 6311963 [PubMed - indexed for MEDLINE]

Publication Types, MeSH Terms

10. Hum Genet. 1981;57(3):300-6.

Aniridia, male pseudohermaphroditism, gonadoblastoma, mental retardation, and del 11p13.

Turleau C, de Grouchy J, Dufier JL, Phuc LH, Schmelck PH, Rappaport R, Nihoul-Fékété C, Diebold N.

Abstract

A 20-month-old male patient was referred because of severe growth and mental retardation, bilateral glaucoma, hypospadias, and cryptorchidism. Karyotyping revealed a de novo complex three-chromosome rearrangement as well as deletion of band 11p13:46,XY,t(4;7;15)(q212;p14;q26)del(11)(p13p14). Trabeculectomia revealed bilateral aniridia. Surgery on the genitalia revealed male pseudohermaphroditism and bilateral gonadoblastoma. The kidneys were normal. A deficiency in catalase (CAT) activity allowed the regional assignment of the CAT gene to band 11p13.

PMID: 6114032 [PubMed - indexed for MEDLINE]

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1. Successful treatment of Fusarium solani ecthyma gangrenosum in a patient affected by leukocyte adhesion deficiency type 1 with granulocytes transfusions.
Mellouli F, Ksouri H, Barbouche R, Maamer M, Hamed LB, Hmida S, Hassen AB, Béjaoui M.
BMC Dermatol. 2010 Oct 7;10:10.
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2. Delayed wound healing in leukocyte adhesion deficiency type 1.
Wada T, Tone Y, Shibata F, Toma T, Yachie A.
J Pediatr. 2011 Feb;158(2):342. Epub 2010 Sep 16. No abstract available.
PMID: 20843524 [PubMed - indexed for MEDLINE]

3. Pyoderma gangrenosum after bone marrow transplantation for leukocyte adhesion deficiency type 1.
Elenberg Y, Shani-Adir A, Hecht Y, Ephros M, Bibi H.
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4. Characterization of 11 new cases of leukocyte adhesion deficiency type 1 with seven novel mutations in the ITGB2 gene.
Parvaneh N, Mamishi S, Rezaei A, Rezaei N, Tamizifar B, Parvaneh L, Sherkat R, Ghalehbaghi B, Kashef S, Chavoshzadeh Z, Isaeian A, Ashrafi F, Aghamohammadi A.
J Clin Immunol. 2010 Sep;30(5):756-60. Epub 2010 Jun 12.
PMID: 20549317 [PubMed - indexed for MEDLINE]

5. A novel point mutation in CD18 causing leukocyte adhesion deficiency in a Chinese patient.
Li L, Jin YY, Cao RM, Chen TX.
Chin Med J (Engl). 2010 May 20;123(10):1278-82.
PMID: 20529581 [PubMed - indexed for MEDLINE] **Free Article**

6. High-dose continuous infusion beta-lactam antibiotics for the treatment of resistant Pseudomonas aeruginosa infections in immunocompromised patients.
Moriyama B, Henning SA, Childs R, Holland SM, Anderson VL, Morris JC, Wilson WH, Drusano GL, Walsh TJ.
Ann Pharmacother. 2010 May;44(5):929-35. Epub 2010 Apr 6.
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7. Osteomyelitis in leukocyte adhesion deficiency type 1 syndrome.
Jabbari Azad F, Ardalan M, Hoseinpoor Rafati A, Sotoudeh S, Pourpak Z.
J Infect Dev Ctries. 2010 Mar 29;4(3):175-8.
PMID: 20351460 [PubMed - indexed for MEDLINE] **Free Article**

8. Leukocyte adhesion deficiency type 1 presenting as leukemoid reaction.
Alizadeh P, Rahbarimanesh AA, Bahram MG, Salmasian H.
Indian J Pediatr. 2007 Dec;74(12):1121-3.
PMID: 18174651 [PubMed - indexed for MEDLINE]

9. Leukocyte adhesion deficiency type 1: an important consideration in the clinical differential diagnosis of prepubertal periodontitis. A case report and review of the literature.
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PMID: 17618138 [PubMed - indexed for MEDLINE]

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J Paediatr Child Health. 2007 May;43(5):406-8.
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13. Retroviral-mediated gene transfer of the leukocyte integrin CD18 into peripheral blood CD34+ cells derived from a patient with leukocyte adhesion deficiency type 1.
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Biochem Biophys Res Commun. 1993 Jun 30;193(3):912-8.
PMID: 7686755 [PubMed - indexed for MEDLINE]

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Toomarian L, Hashemi N.
Arch Iran Med. 2010 Jul;13(4):355-9.
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9. Leukocyte adhesion deficiency type II: long-term follow-up and review of the literature.
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11. Chronic eczema in a patient with Leukocyte Adhesion Deficiency (LAD) type I.
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A family with Papillon-Lefevre syndrome reveals a requirement for cathepsin C in granzyme B activation and NK cell cytolytic activity.

Meade JL, de Wynter EA, Brett P, Sharif SM, Woods CG, Markham AF, Cook GP.

Leeds Institute of Molecular Medicine, University of Leeds, St James's University Hospital, Leeds LS9 7TF, United Kingdom.

Abstract

Activation of granzyme B, a key cytolytic effector molecule of natural killer (NK) cells, requires removal of an N-terminal pro-domain. In mice, cathepsin C is required for granzyme processing and normal NK cell cytolytic function, whereas in patients with Papillon-Lefèvre syndrome (PLS), loss-of-function mutations in cathepsin C do not affect lymphokine activated killer (LAK) cell function. Here we demonstrate that resting PLS NK cells do have a cytolytic defect and fail to induce the caspase cascade in target cells. NK cells from these patients contain inactive granzyme B, indicating that cathepsin C is required for granzyme B activation in unstimulated human NK cells. However, in vitro activation of PLS NK cells with interleukin-2 restores cytolytic function and granzyme B activity by a cathepsin C-independent mechanism. This is the first documented example of a human mutation affecting granzyme B activity and highlights the importance of cathepsin C in human NK cell function.

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Diagnosis and carrier detection of chronic granulomatous disease in five families by flow cytometry.

Crockard AD, Thompson JM, Boyd NA, Haughton DJ, McCluskey DR, Turner CP.

Regional Immunology Laboratory, Royal Victoria Hospital, UK. a.crockard@qub.ac.uk

Abstract

BACKGROUND: The application of flow cytometric assays, for determination of phagocyte respiratory burst (ROB) activity, to the investigation of chronic granulomatous disease (CGD) may lead to improved laboratory detection of patients and carriers and indicate the nature of the molecular defect. To evaluate the diagnostic capability of flow cytometry an investigation of 5 CGD families was undertaken.

METHODS: Phorbol myristate acetate (PMA)-induced neutrophil ROB was determined using dihydrorhodamine 123 (DHR) and flow cytometric analysis in 26 members of 5 CGD families (2: X-CGD; 3: autosomal recessive CGD).

RESULTS: Neutrophils from X-CGD patients displayed absence of reactivity. Female carriers demonstrated dual fluorescence peaks of high and low intensity indicative of normal and abnormal populations, respectively. Normal ROB activity was observed in a boy whose X-CGD was successfully treated by bone marrow transplantion. Reduced ROB activity was observed in 3 patients with autosomal-recessive CGD compared with their parents and siblings. The patterns of flow cytometric reactivity correlated with the different molecular defects identified. Absence of the p22phox membrane component of the NADPH oxidase complex resulted in a significantly reduced level of respiratory burst activity which was comparable to that observed in X-CGD, whereas reduced but detectable levels of respiratory burst activity were observed in a patient with diminished levels of p22phox and in a patient with deficiency of the cytosolic p47phox component.

CONCLUSIONS: The DHR flow cytometric assay offers a sensitive diagnostic screening test for CGD and furthermore may provide an indication of the likely underlying molecular defect.

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Molecular characterization of autosomal recessive chronic granulomatous disease caused by a defect of the nicotinamide adenine dinucleotide phosphate (reduced form) oxidase component p67-phox.

Patiño PJ, Rae J, Noack D, Erickson R, Ding J, de Olarte DG, Curnutte JT.

Department of Immunology, Genentech, Inc, South San Francisco, CA, USA.

Abstract

Chronic granulomatous disease (CGD) is a rare inherited disorder of phagocytes in which defective production of microbicidal oxidants leads to severe recurrent infections. CGD is caused by mutations in any of 4 genes encoding components of nicotinamide adenine dinucleotide phosphate (reduced form; NADPH) oxidase, the multisubunit enzyme that produces the precursor of these oxidants, superoxide. Approximately 5% of CGD patients have an autosomal recessive form of disease caused by a severe deficiency of p67-phox, a 526-amino acid subunit of the oxidase that appears to regulate electron transport within the enzyme. Here we report the biochemical and molecular characterization of 6 unrelated kindreds with p67-phox deficiency. These studies show that, as in gp91-phox and p22-phox deficiencies, the p67-phox CGD patients show a high degree of heterogeneity in the genetic defects that underlie their disease. Five different mutant alleles were identified: (1) a nonsense mutation in exon 4 (C(304) --> T); (2) a 5-nucleotide (nt) deletion in exon 13 (nts 1169-1173); (3) a splice mutation in the first nucleotide of intron 4 (G --> A); (4) a deletion of 1 nt in exon 9 (A(728)); and (5) a 9-nt in-frame deletion in exon 2 (nts 55-63). The splice mutation was seen in 3 unrelated kindreds, while the 5-nt deletion was seen in 2 apparently unrelated families (both of Palestinian origin). Homozygosity was present in 4 of the kindreds, 2 of which had consanguineous parentage. In the isolated neutrophils of each of the affected patients in the 6 kindreds, there was no measurable respiratory burst activity and no p67-phox protein detected by immunoblot analysis. The level of 67-phox mRNA was less than 10% of normal in the mononuclear leukocytes from 3 of the 4 patients analyzed by Northern blot studies. Thus, this heterogeneous group of mutations in p67-phox all lead to marked instability of mRNA or protein (or both) that results in the complete loss of NADPH oxidase activity.

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1. Chronic granulomatous disease caused by mutations other than the common GT deletion in NCF1, the gene encoding the p47phox component of the phagocyte NADPH oxidase.
   Roos D, de Boer M, Köker MY, Dekker J, Singh-Gupta V, Ahlin A, Palmblad J, Sanal O, Kurenko-Deptuch M, Jolles S, Wolach B.
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Clin Immunol. 2008 Aug;128(2):117-26.

Focus on FOCIS: the continuing diagnostic challenge of autosomal recessive chronic granulomatous disease.

Yu G, Hong DK, Dionis KY, Rae J, Heyworth PG, Curnutte JT, Lewis DB.

Division of Immunology and Transplantation Biology, Department of Pediatrics, Stanford University School of Medicine, Stanford, CA 94305, USA.

Abstract

Chronic granulomatous disease (CGD) is a primary immunodeficiency of defective neutrophil oxidative burst activity due to mutations in the genes CYBA, NCF-1, NCF-2, and CYBB, which respectively encode the p22-phox, p47-phox, p67-phox, and gp91-phox subunits. CGD usually presents in early childhood with recurrent or severe infection with catalase-positive bacteria and fungi. We present an unusual case of CGD in which Burkholderia cepacia lymphadenitis developed in a previously healthy 10-year-old girl. Flow cytometric analysis of dihydrorhodamine (DHR)-labeled neutrophils performed by a CLIA-approved outside reference laboratory was reported as normal. However, we found that this patient's neutrophil oxidative burst activity in DHR assays was substantially reduced but not absent. A selective decrease in intracellular staining for p67-phox suggested the diagnosis of autosomal recessive CGD due to NCF-2 gene mutations, and a novel homozygous and hypomorphic NCF-2 gene mutation was found. The potential mechanisms for this delayed and mild presentation of CGD are discussed.

PMID: 18625437 [PubMed - indexed for MEDLINE]

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