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Infections and Inborn Errors of Immunity in Children

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Abstract

Introduction: Inborn errors of immunity (IE) are rare genetic disorders that cause immune dysfunction. In Africa, they are underdiagnosed due to a lack of awareness, the scarcity of specialists in this field, and the difficulty of diagnosis. They are associated with atypical infections, autoinflammation, autoimmunity, and neoplasms. In children, infections are often the presenting symptoms or a concerning complication due to their severity and frequent recurrence. Our objective was to describe the profile of these infections in children with IE followed in a pediatric hospital. **Patients and Methods:** We conducted a retrospective descriptive and analytical study of children followed for inborn errors of immunity. We included those who followed from February 2016 to September 2025, aged under 16 years, and who had experienced at least one infectious episode. Sociodemographic, clinical, biological, microbiological, and outcome data were collected from medical records and analyzed using SPSS 27. **Results:** Of the 44 children followed for inborn errors of immunity (IE) during our study period, 33 had experienced at least one infectious episode, representing a frequency of 75%. However, infection was documented in 25 children. The mean age of the children was 9 ± 5 years, with a slight female predominance (51.5%). A total of 140 infectious episodes were recorded, with the most frequent sites being cutaneous and pulmonary. Cytobacteriological examination of pus was the most frequently requested microbiological analysis (22.6%). The main pathogens isolated were Staphylococcus aureus, Mycobacterium tuberculosis, Candida albicans, Candida tropicalis, and

Trichophyton rubrum. The most common inborn errors of immunity that cause infections were Wiskott-Aldrich syndrome, hyper-IgE syndrome (HIES), and epidermodysplasia verruciformis. Anti-infective treatment consisted primarily of antibiotics and antifungals. Prevention of recurrent infections was achieved with cotrimoxazole prophylaxis. **Conclusion:** In our study, skin and respiratory infections were the most frequent. Their origin was primarily fungal and bacterial. The most common inborn errors of immunity were Wiskott-Aldrich syndrome, hyper-IgE syndrome, and epidermodysplasia verruciformis, hence the need for their prevention.

Keywords

Infections, Children, Inborn Errors of Immunity (IEI)

1. Introduction

Inborn errors of immunity (IEI) constitute a heterogeneous group of approximately 500 rare syndromes resulting from genetic disorders affecting the functioning of the immune system. The global prevalence is estimated at 1 in 5000 births [1]. According to the 2021 report by the Jeffrey Modell Foundation (JMF), 258,392 patients with IEI are being monitored worldwide, with 79,638 cases reported in the United States, 7500 in France, and 2936 in Africa [2]. Despite scientific advances, IEI remains underdiagnosed, particularly in resource-limited countries, where its recognition and management still pose numerous challenges. The main manifestations are predispositions to infections, neoplasms, or autoimmunity due to immune dysfunction of genetic origin. Indeed, infections are their primary manifestation in children. Analyzing their profiles helps guide diagnosis and offer early intervention to prevent the risk of complications. Our objective was to study the epidemiological, clinical, biological, and microbiological characteristics of these infections in children monitored for inborn errors of immunity.

2. Patients and Methods

This was a retrospective, cross-sectional, descriptive, and analytical study conducted at the Outpatient Unit for Children and Adolescents with Sickle Cell Disease (USAD) of the Albert Royer National Children's Hospital (CHNEAR) in Dakar, involving children followed from February 2016 to September 2025. Those under 16 years of age who had experienced at least one infectious syndrome during follow-up were included. We excluded those with incomplete or unusable information. Data were collected using an Excel survey form, from the database, and from patients' medical records. Statistical analysis was performed using IBM SPSS 27 software, and graphs were generated with Microsoft Excel. Quantitative variables were expressed as counts, means with standard deviation, and median with ranges. Qualitative variables were presented as counts and percentages.

3. Results

During our study period, we identified 44 children with inborn errors of immunity (IEI). Among them, 33 had experienced at least one infectious episode, representing a frequency of 75%. A slight female predominance was observed (17/33 girls (51.5%), for a sex ratio of 0.94). The mean age at the time of the study was 103 ± 58 months (9 ± 5 years), with a median of 88 months (7 years) and a range from 23 months to 213 months (18 years). Parental consanguinity was reported in 13/33 children (39.4%). A personal history of neonatal infections was found in 5/33 children (15.2%), and a family history of inborn errors of immunity was reported in 4/33 children (12.1%). The most frequently observed type of infection was pneumonia (15.2%). The distribution of infectious episodes during the follow-up years showed a peak in 2019 and 2021, separated by a decrease case in 2020 (Figure 1).

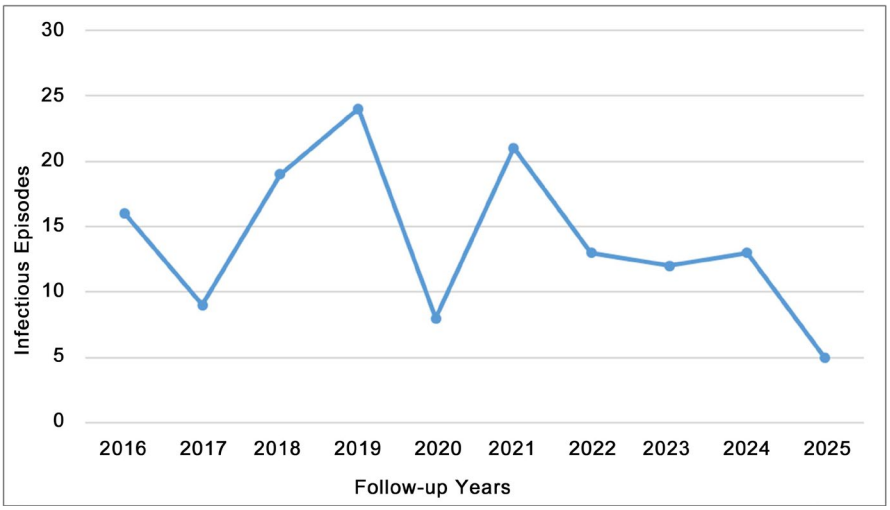

Figure 1. Distribution of infectious episodes during follow-up.

Clinically, the main manifestations suggestive of inborn errors of immunity were infectious, and the most frequent locations were cutaneous and pulmonary (Table 1).

Table 1. Characteristics of infections according to site, type, and frequency.

Site	Type of infectious manifestations	Frequency (Episodes number)
Oral		**13**
	Thrush	9
	Stomatitis	1
	Dental caries dentaires	3
Cutaneous		**42**
	Impetigo	8

Continued

	Tyrcosis	5
	Cut Abscess	5
	Tineum Scalp	4
	Other fungal infections	4
	Unspecified dermatoses	4
	Adenitis	3
	Warts	3
	Grenoble Creutzfeldt-Eye Rash	3
	Molluscum contagiosum	2
	BCGitis	2
	Superinfected Eczema	1
Pulmonary		**30**
	Pneumonia	14
	Bronchitis	9
	Bronchopneumonia	3
	Tuberculosis	3
	Lung Abscess	1
ENT		**21**
	Tonsillitis	7
	Otitis	6
	Rhinorrhea	8
Digestive		**10**
	Recurrent diarrhea	4
	Acute Gastroenteritis (AGE)	6
Neurological		**2**
	Meningitis	1
	Encephalitis	1
Ocular		**2**
	Conjonctivitis	2
Uro-genital		**1**
	Orchitis/Epididymitis	1

A microbiological study was conducted on 16 children, with a total of 62 samples, primarily cytobacteriological examinations (Table 2).

Table 2. Types and number of microbiological samples.

Sample Type	Number (n)	Percentage (%)
Blood	18	29.0
Pus	14	22.6
Stolls	5	8.1
Urine	5	8.1
CSF	4	6.5
Gastric tube	4	6.5
Sputum	3	4.8
Skin	3	4.8
Sputum	2	3.2
Puncture fluid	2	3.2
Scalp	1	1.6
Pleural fluid	1	1.6
Total	**62**	**100.0**

A total of 23 pathogens were identified, with Staphylococcus aureus being the most prevalent (n = 7, 30.4% of isolated organisms), followed by Candida albicans and Mycobacterium tuberculosis (n = 3 each, 13.0%) (Table 3).

Table 3. Types and numbers of isolated pathogens.

Category	Pathogen	Number (n)	Percentage (%)
Bacteria		**18**	**78.3**
	Staphylococcus aureus	7	30.4
	Mycobacterium tuberculosis (BK)	3	13.0
	Proteus mirabilis	2	8.7
	Methicillin-susceptible Staphylococcus aureus	2	8.7
	Gram-negative non-fragmented bacillis	1	4.3
	Enterobacter spp	1	4.3
	Pseudomonas aeruginosa	1	4.3
	Streptococcus pneumoniae	1	4.3
Fungi		**5**	**21.7**
	Candida albicans	3	13.0
	Candida tropicalis	1	4.3
	Trichophyton rubrum	1	4.3
General total		**23**	**100.0**

Respiratory infections were the most frequent, and Wiskott-Aldrich syndrome was the leading cause of infection (Table 4).

Table 4. Classification of isolated pathogens according to the site of infection and type of EII.

Localization	Pathogens	Suspected Inborn Error of Immunity
Respiratory	*Streptococcus pneumoniae*, *Mycobacterium tuberculosis*, *Candida albicans*, *Candida tropicalis*	**WAS**, **HIES**, **Verruciform epidermodysplasia**
ENT	*Pseudomonas aeruginosa*, *Staphylococcus aureus*, *Proteus mirabilis*, *Enterobacter spp*, *Candida albicans*	**WAS**, **CMH-II**, **PI3K Deficiency**
Cutaneous	*Staphylococcus aureus*, *Candida albicans*, *Trichophyton rubrum*, *Mycobacterium tuberculosis*	**GSC**, **WAS**, **EV**, **MSMD, cyclical Neutropenia**,
Digestive	*Candida tropicalis*	**CMC**
Neurological	*Proteus mirabilis*	**Hypogammaglobulinemia**
Lymph node	*Mycobacterium tuberculosis*	**GSC**, **SCID**
Oral/Ocular	*Staphylococcus aureus*	**WAS**

Analysis of antibiograms showed 100% bacterial susceptibility to the majority of first-line antibiotics, such as amoxicillin, ceftriaxone, ciprofloxacin, and vancomycin. Genetic diagnosis was confirmed in 5/33 children (15.2%) by molecular testing.

Regarding management, of the 140 documented infectious episodes, 39 (27.9%) required hospitalization, with an average duration of 14.4 days and a range from 1 to 103 days (3.5 months). Antifungals were the most frequently prescribed class of medication (n = 22), followed by antibiotics and antiparasitics.

4. Discussion

Given the size of the Senegalese population, this sample corresponds to an extremely low frequency. This constitutes a limitation for the study. Indeed, in other countries such as the United States and some pioneering European countries, 158 cases had already been reported by 1995 [3]. This difference could be explained by a lack of awareness of these conditions, the scarcity of specialists in this field, and the difficulties in diagnosis. They are underdiagnosed due to their wide range of presentations. The distribution according to sex varies from one series to another and according to the mode of transmission of the condition, as evidenced by data from the literature, in Senegal and elsewhere in Africa and the world [4]-[6]. We found a slight female predominance; however, a male predominance is

usually observed in disorders with X-linked deficiencies, such as some forms of severe combined immunodeficiency (SCID) [7]. The mean age of the patients was 49.94 months (4 years and 2 months), with a median of 28 months, reflecting considerable variability.

in the age at diagnosis. Indeed, the majority of children (42.4%) were under 2 years old at enrollment, reflecting the early severity of some forms, particularly severe combined deficiencies or antibody deficiencies. Similar results were reported by Kane *et al.* in 2021 on this same cohort and in a Jordanian study from 2024 [8]. In contrast, in a multicenter pediatric study conducted in India in 2024, Sabui *et al.* reported a younger age, with a median of 8 months (4 - 24 months) [9].

Indeed, inborn errors of immunity can occur at any age, but are generally present at birth. This can involve deficiencies in non-specific or innate immunity (phagocytes, complement) or deficiencies in specific or adaptive immunity (humoral, cellular, or combined immunodeficiencies).

In our cohort, 5 patients (15.15%) had a history of neonatal infections and 16 (48.48%) had a history of post-neonatal infections. These results demonstrate the close link between invasive encephalopathy and increased vulnerability to infections and are comparable to those reported in the literature. Indeed, a multicenter study in India showed that all children with IE had a history of recurrent infections [6]. Another study reported that 59.5% of children hospitalized for severe infections were infants [10]. A history of parental consanguinity was found in 13/33 children (39.4%). These figures, although high, are considerably lower than those described. in regions with high rates of endogamy. Indeed, North African studies reported much higher rates of consanguinity, between 61.1% and 71% [5] [11]. These results confirm the role of consanguinity as a risk factor that promotes the expression of autosomal recessive mutations, frequently implicated in severe forms of immune dysregulation (IDD), particularly severe combined immunodeficiencies. In our cohort, the distribution of inborn errors of immunity according to the IUIS classification showed a predominance of phagocytic deficiencies. Syndromic and antibody deficiencies were the second most common. Combined and complement deficiencies were less frequent.

Infectious Manifestations during Follow-up

In our study, infections were the main warning signs for suspecting an innate error of immunity (IE). They were varied, reflecting the heterogeneity of the clinical presentations of these conditions. The most frequent infections were pulmonary in origin, predominantly pneumonia. A single-center study conducted in Jordan reported 55.2% pulmonary involvement [8]. These results confirm the particular vulnerability of patients with IE to respiratory infections. ENT infections were also frequent, including rhinorrhea, tonsillitis, and otitis media [12]. Cutaneously, we observed a significant frequency of impetigo, tinea corporis, and skin abscesses. These dermatological manifestations are classically observed in the form of recurrent skin infections [13].

Other infections were also observed, such as stomatitis, epididymitis/orchiitis, ocular or urogenital infections, and chronic diarrhea [8] [12]. Indeed, the close link between infections and genetic predisposition was highlighted starting in the 1940s, with the emergence of the genetic theory, which complemented the microbial theory. The latter made it possible to understand that the occurrence of severe or recurrent infections depends not only on the presence of the microbe, but also on genetic defects that impair the functioning of the immune system [14]. However, the occurrence of these infections is influenced by environmental factors. Indeed, during the COVID-19 pandemic, the peak incidence of infections decreased, demonstrating the effectiveness of individual and collective preventive measures during this period (2020-2021). We observed that 75% of hospitalized children presented with combined immunodeficiency (CID), reflecting the increased vulnerability and clinical severity of this form compared to other types of deficiencies.

This result contrasts with several pediatric studies that reported a predominance of humoral deficiencies. In fact, an initial study conducted on children aged 0 to 12 years found 37% with humoral deficiencies, involving 81 children hospitalized for recurrent infections, reported an even higher frequency of 36.7% [9]. Elsewhere, other studies showed other results [15]. This difference could be explained by recruitment biases and the limited size of our sample, but also by the greater severity and more pronounced clinical presentation of combined forms in our population, facilitating their identification in the hospital. The mean length of hospital stay was 14.4 days (median: 7 days, range: 1 - 103). This duration is longer than that reported in the ARS 2024 registry (7.4 days) [16] as well as that observed in the United States (5.6 days) [17]. This difference could be explained by the predominance of combined immunodeficiency (CID) in our cohort, the increased severity of opportunistic infections, and the diagnostic and therapeutic delays often encountered in resource-limited settings.

From a biological standpoint, anemia was found in 71.4% of blood counts; it was predominantly hypochromic microcytic [18]. Thrombocytopenia was also frequent (59.6%) in Wiskott-Aldrich syndrome and MHC deficiency. The high frequency of leukopenia (18.4%) and especially neutropenia underscores the intrinsic immune vulnerability of these conditions, predisposing individuals to severe bacterial and fungal infections, which explains the infections in our population. Thus, this alternation between leukocytosis (response) and cytopenias (deficiency) illustrates the unbalanced struggle between infection and immune defense in immunocompromised children [19]. A total of 23 pathogens were isolated, with a clear predominance of bacteria. Indeed, a similar result was also reported in the Algerian registry of immune system infections (ISI), with 557 pathogens identified and a bacterial predominance exceeding 60% [20]. Another Chinese study also showed a predominance of bacterial agents (91.52%). In contrast, no viruses were isolated in our cohort, contrary to data in the literature [21]. This could be explained by the absence of systematic virological testing, apart from retroviral se-

rology performed to rule out HIV infection and support the hypothesis of an inborn error of immunity. In our cohort, several children with combined immunodeficiencies had developed severe bacterial infections. In a child with Wiskott-Aldrich syndrome, Staphylococcus aureus was isolated from several infectious foci, including skin abscesses, otitis media, stomatitis, and conjunctivitis. Mycobacterium tuberculosis was responsible for pulmonary tuberculosis in the same child. This extensive clinical presentation illustrates the mucocutaneous fragility characteristic of this severe combined deficiency [22]-[24]. Another child with PI3K deficiency had developed recurrent otitis media due to Pseudomonas aeruginosa, S. aureus, and Enterobacter spp., while another child with MHC II deficiency had experienced otitis media with Proteus mirabilis. Furthermore, a child with HyperIgE syndrome developed a Streptococcus pneumoniae lung abscess.

Indeed, HyperIgE syndrome is classically associated with infections. Severe and recurrent bacterial infections, such as Staphylococcus aureus, preferentially affect the skin, lungs, and deep tissues [11]. Overall, our results were comparable to data in the literature, confirming that children with combined immunodeficiency (CID) have increased susceptibility to bacterial infections, characterized by their frequency, early onset, severity, and polymorphism. The main pathogens associated with CID isolated in our cohort are also well documented in the literature:

- Staphylococcus aureus: responsible for skin and lung abscesses, osteomyelitis, pneumonia, septicemia, impetigo, and vesicular skin lesions.
- Streptococcus pneumoniae: implicated in pneumonia, otitis, sinusitis, sinopulmonary infections, meningitis, and septicemia.
- Pseudomonas aeruginosa: found in pneumonia, enterocolitis, and cholangitis, particularly in patients with chronic lung disease [25]. We observed a case of BCG infection due to Mycobacterium tuberculosis in a child with an innate immune deficiency, specifically Mendelian Susceptibility to Mycobacterial Disease (MSMD) linked to a STAT1 gene mutation. This case typically illustrates the increased vulnerability to mycobacterial infections in patients with innate immune deficiencies, particularly those affecting the IFN-γ/IL-12 signaling pathway. This pathway plays a central role in macrophage activation, which is essential for containing intracellular infections. In cases of failure, such as in STAT1 mutations, patients become particularly susceptible to low-virulence mycobacteria, including BCG. BCGitis thus appears as a major clinical marker in innate immunity deficiencies and should raise suspicion of MSMD, especially when it occurs early, after vaccination [24]. In patients with combined immunodeficiency disorders (CID), particularly Wiskott-Aldrich syndrome and PI3K deficiency, fungal infections primarily affected the respiratory tract and the ENT region. Indeed, susceptibility to fungal infections in CID is mainly explained by impaired adaptive immunity. These patients exhibit abnormalities of T and B lymphocytes, leading to a failure of the cellular and humoral response necessary for controlling opportunistic fungi, such as Candida spp. Regarding innate immunity deficiencies, one case involved a

child with chronic mucocutaneous candidiasis (CMC) in whom a Candida tropicalis infection was identified, with simultaneous gastrointestinal and pulmonary involvement and a positive blood culture, indicating systemic candidiasis. Indeed, CMC is a rare disease, linked to genetic abnormalities (altering the IL-17/Th17 pathway, essential in antifungal defense [26], was also identified. Another case of papillomavirus infection was diagnosed in a child with epidermodysplasia verruciformis (EV), a rare genetic disorder classically associated with increased susceptibility to papillomavirus skin infections.

This situation illustrates how certain genetic conditions can create a favorable environment for cross-infections, particularly fungal infections, in the context of inborn errors of immunity [24]. These results are comparable to data in the literature, which clearly demonstrate the increased sensitivity of innate immunity deficiencies to fungal infections, especially Candida spp. and other opportunistic fungi. Indeed, innate immunity plays a central role in the recognition and control of fungi via key pathways such as Toll-like receptors (TLRs), the IL-17/Th17 pathway, and the IFN-γ/IL-12 axis. When these pathways are impaired, as in CMC, patients develop a marked susceptibility to cutaneous, mucosal, or deep fungal infections, thus confirming the role of fungi as major pathogens in inborn errors of immunity [27].

Radiographically, pneumonias represented the majority of respiratory abnormalities (46.6%) and primarily affected patients with humoral deficiencies and combined immunodeficiencies (SCID, PI3K deficiency, MHC class II deficiency, Hyper IgE syndrome). Recurrent pneumonias pose a risk of long-term complications such as bronchiectasis, identified in three patients, including those with Wiskott-Aldrich syndrome, SCID, and epidermodysplasia verruciformis. Antibiotic prophylaxis was administered to more than half of the patients (54.54%), primarily with cotrimoxazole (Bactrim). This approach aligns with international recommendations, which consider antibiotic prophylaxis an essential pillar in preventing recurrent bacterial infections and reducing morbidity and mortality in patients with invasive fungal infections (IFI) [5] [6] [8] [28]. Conversely, the low rate of antifungal prophylaxis (5% of patients) could be explained by its unavailability and inaccessibility.

None of our patients had received immunoglobulin replacement therapy or hematopoietic stem cell transplantation (HSCT), reflecting the limited access to these specific treatments in our setting. This situation contrasts with data reported in other countries: in Jordan, 63% of patients received IVIG replacement therapy and 30.8% underwent transplantation [29], while in China, 39.83% of patients received transplantation [29]. According to the European registry of the Jeffrey Modell Foundation (JMF), 141,142 patients with primary immunodeficiency were identified, of whom only 2703 had received a hematopoietic stem cell transplant (HSCT), a relatively low rate. Across the African continent, the same JMF study reported 70 transplants out of 2936 patients, illustrating the rarity of this therapeutic option, except in North African countries [27]. During the follow-up pe-

riod, 64% of patients experienced recurrent infections. Other complications observed were primarily bronchiectasis (BD) and sepsis, reflecting the natural progression of severe combined immunodeficiency (SCID) or untreated humoral immunodeficiency [29]. The observed mortality rate was 18.2%, with SCID accounting for 50% of cases. Infections were the leading cause of death. In Jordan, the overall mortality rate was 33.2%, with the highest rate observed in severe combined immunodeficiency (SCID: 56.2%) [6]. This underscores the severity of SCID and the need for early diagnosis, appropriate management, and close monitoring to improve prognosis.

In recent years, the identification of molecular abnormalities has significantly improved, thanks to training and advances in genetic sequencing. However, in our study, only 5/33 children (15.2%) had genetic confirmation of their diagnosis by molecular biology. This low rate of genetic confirmation reflects the limited access to molecular biology in our setting. It contrasts sharply with data reported in other countries. Al-Herz *et al.* in Kuwait reported a rate of genetic testing performed in 69% of patients, with a confirmation rate reaching 90% [29]. A study conducted in southern India reported the systematic performance of genetic testing in all suspected cases of IBD, representing nearly 205 children [29]. Thus, the number of genes involved has increased from around one hundred in the 2000s to more than 450 to date [29]. In total, 140 infectious episodes were recorded in 24 children over a ten-year period, representing an average of approximately 5.8 episodes per patient. Among them, 12 children (36.4%) required at least one hospitalization. This illustrates the high infectious burden observed in children with IBD.

5. Conclusions

In our study, inborn errors of immunity were characterized by a marked susceptibility to infections. The infectious sites were primarily pulmonary or cutaneous. Their origins were often fungal or bacterial.

The course of the illness was marked by relapses and severe complications such as bronchiectasis.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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