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Inborn Errors of Immunity with Invasive Fungal Infections: A Single Center Experience

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Abstract

Purpose: Data about invasive fungal infections in patients with inborn errors of immunity (IEI) in Egypt are still inadequate.

Methods: We herein describe fungal infections encountered in a group of infants and children with IEI.

Results: Out of 680 patients with IEIs, 42 patients had documented invasive fungal infections. They were 26 boys (62%) and 16 girls (38%) with age ranging 0.3-13 years, median 3.5 years. According to their diagnoses, 15 had chronic granulomatous disease (35.7%), 6 severe combined immunodeficiency (14.3%), 3 LRBA mutation (7%), 2 CD3 lymphopenia (4.7%), 2 congenital neutropenia (JAGN1 mutation), one autoimmune lymphoproliferative syndrome and one with each of the following mutations: STAT3 GOF, STAT 1 GOF, CARD 9, CARD 11, TRNT1, DOCK8 and PIK3C2G. The remaining 6 patients are still under investigation. Mucocutaneous infections were the most common (22, 52.4%) followed by pulmonary affection (19, 45.2%), bone/joint lesions (8, 19%), intracranial lesions (7, 16.7%), hepatic lesions (4, 9.5%), eye invasion (3, 7%) and intracardiac fungal masses in 2 (4.7%). Aspergillus and candida species were equally prevalent among the described cases (each in 21 patients; 50%), followed by pneumocystis jirovecii (n=3, 7%), Mucormycosis in one and Exophilia dermatitidis in one. Antifungal therapy included voriconazole in 31 (73.8%), amphotericin in 24 (57%), fluconazole and posaconazole, each in 9 (21.4%), and anidulafungin in 2 (4.7%), while 14 patients (33.3%) required combined antifungal therapy. The mortality rate among our patients reached 23/42 (54.8%).

Conclusion: Timely diagnosis of fungal infections in patients with IEI and antifungal availability are still challenging.