Phenotypic expressions of light combined immunodeficiency (cellular or humoral): report of three cases.

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Introduction

The primary immunodeficiency represents more them 130 different diseases that affect the immune function and, therefore, the development of the children. Although they are simple monogenic diseases, the variability of expression comes from many different interactions between genetic and environmental factors.[1,2]

The aim of this study was to report three cases of light combined immunodeficiency, which showed distinct phenotypic expressions, and to analyze the possible correlated genetic mutations associated with the manifestations described in these cases.

Cases

Patient 01 – 1 year, male, had frequent hospitalizations for recurrent infections such as pneumonia, infection of urinary tract and sepsis after cellulites, and he had lymphopenia in sequential tests. The patient was wheezing baby whose need medications for control. The immunoglobulins presents in 97th percentile, CD3 less than 50th percentile, CD4 and CD8 in 50th percentile and CD19 less than 10th percentile. The treatment was immunoglobulin, a total of six dose, with general improvement.

Patient 02 – 7 years, male, had suffered from allergic rhinitis in regular use of medications and he'd showed lymphopenia. No history of infections requiring intervention. The immunoglobulins presents between percentile 50th and 75th. The levels of CD4, CD8, CD16, CD19, CD20 and CD 56 in percentile less than 10th. The treatment was prophylactic use of antibiotic (azithromycin) and regular use of medications for the control of rhinitis.

Values of immunoglobulins, lymphocytes and complement

	Patient 01	Patient 02	Patient 03
Immunoglobulins Ig G Ig M Ig A Ig E	 1100 (P97) 148 (P97) 85 (P50) 24.2 (P50)	 1350 (P75) 110 (P50) 168 (P50) 61 (P50)	 1426 (P75) 143 (P75) 239 (P97) 141 (P50)
CD3	3749 (>P50)	-	-
CD4	19,66 (P50)	447 (<P10)	-
CD8	1485 (P50)	796 (<P10)	-
CD19	243 (<P10)	107 (P<10)	-
CD16,CD20 and CD 56	-,-,-	107, 288, 207 (<P10)	-,-,-
C3, C4	-,-	132, 29	188, 42

Patient 03 – 3 years, female, had repeated infections associated metabolic disease (fructose bifosfatase 1.6 deficiency), and she'd required ICU support. She had neutrophilia. The immunoglobulins presents between percentiles 50th and 97th, CD3 and CD4 in normal values. The treatment was prophylactic use of antibiotic (trimethoprim-sulfamethoxazole), chelated zinc and medication for metabolic disease (carnitine, biotin, folic acid, sodium bicarbonate and captopril).

Discussion

The main clinical manifestation of immunodeficiency diseases is increased susceptibility to infections. The pattern of involvement and the associated pathogens vary with the type of immunity affected..[1,3]

The abnormal development of B cells results in a deficiency in antibody production, increasing susceptibility to infections caused by extracellular microorganisms, particularly capsulated germs. The diagnosis of immunodeficiency diseases associated with B lymphocyte serum has reduced immunoglobulin deficiency in response to vaccination and reduced number of B cells in the circulation or in lymphoid tissues or absence of plasma cells in tissues.[3] Among the variety of disorders associated with B lymphocytes, we have the X-linked agamaglobulinemia, immunoglobulin deficiency, common variable immunodeficiency and hyper-IgM syndrome X-linked.

Abnormalities in T-lymphocyte activation have become more frequent. Among them is the defect in the expression of histocompatibility complex (MHC), with defective expression of MHC II or deficiency in the levels of MHC I. [3]

Conclusion

The main clinical manifestation of immunodeficiency diseases is the increased susceptibility to infections. The pattern of involvement and the associated pathogens varies with the type of immunity affected. In this group there was great heterogeneity of abnormalities: There were defects in expression of CD3 subunits, in the expression of tyrosine kinase ZAP 70, in the synthesis of cytokines, in intracellular calcium influx, in expression of FAS, and other expression's defects. The report of these three cases raises in the issue of the gap between clinical diagnosis and molecular diagnosis. Because the molecular diagnose is expensive and often not feasible, the primary immunodeficiency diagnoses' depends on clinical presentation, without necessarily need for microbiological evidence.

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