Saving one more life?
Easy as reading this leaflet

Discover PI3Kδ Activation Syndrom (APDS)

— 1.What is it?

A rare primitive immunodeficiency, characterized by impairment either concerning the humoral immunity, with agammaglobulinemia of variable severity and IgM increase, or concerning the cellular immunity, that leads to the circulation of immature and aging cells B and T, however not immunocompetent.

— 2.How it looks like

The disease appears since childhood with **recurrent and/or serious bacterial and viral infections** *(specifically bacterial sinopulmonary infections and due to herpes virus)*, **benign chronic lymphoproliferation** *(that starts with lymphadenopathy, hepatosplenomegaly, nodular lymphoid focal hyperplasia and lymphomas)* and/or **autoimmune diseases** *(cytopenias, arthritis, glomerulonephritis and sclerosing cholangitis)*.

— 3.How it develops

This disease is caused by **genetic alterations in the genes PIK3CD and PIK3R1** that encode respectively the activation or the adjustment of PI3Kδ protein. These alternations cause **abnormalities in lymphocytes** T and B operating.

— 4.How to diagnose it

Useful clues to raise the clinical suspicion about APDS are: **frequent occurrence of infections, presence of persistently enlarged lymph nodes, increased size of liver and/or spleen, low levels of IgG and IgA immunoglobulins in presence of high IgM, reduced and all immature B lymphocytes, unbalanced T lymphocytes in comparison with the terminally differentiated ones.** The APDS definite diagnosis is only made by genetic analysis of PIK3CD and PIK3R1 genes encoding the two PI3Kδ subunits.

— 5.How do we cure it?

The only treatment about APDS, in the rare cases of compatibility, is the **Hematopoietic Stem Cell Transplant**. The symptomatic treatment is focused instead on anti-infective and anti-inflammatory drugs, substitutional therapies with immunoglobulins, other immunosuppressant drugs.

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