

The role of the Laboratory in the diagnosis of Primary Immunodeficiency Diseases

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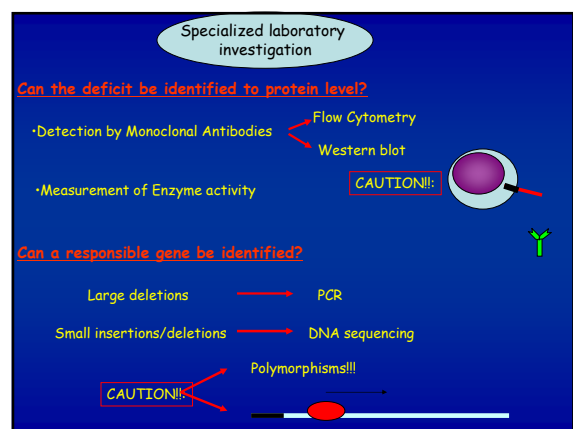
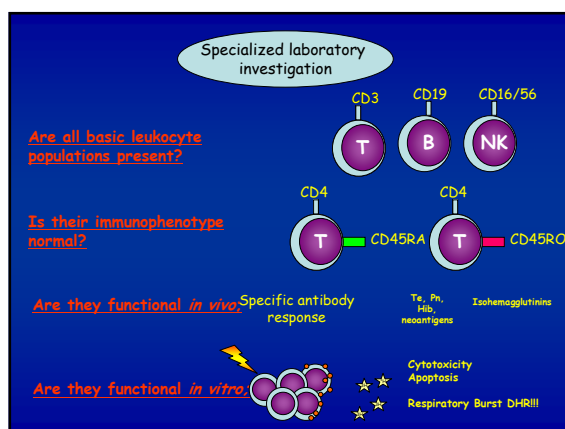
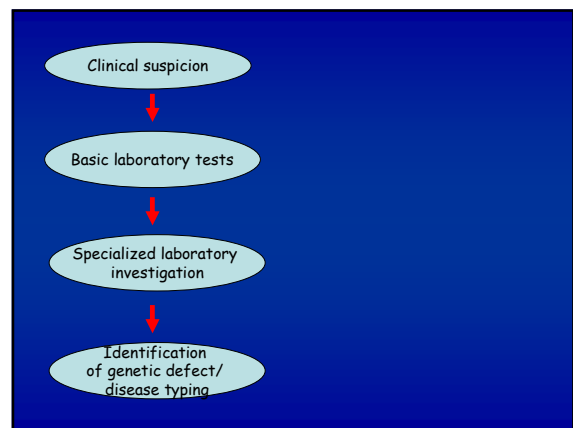
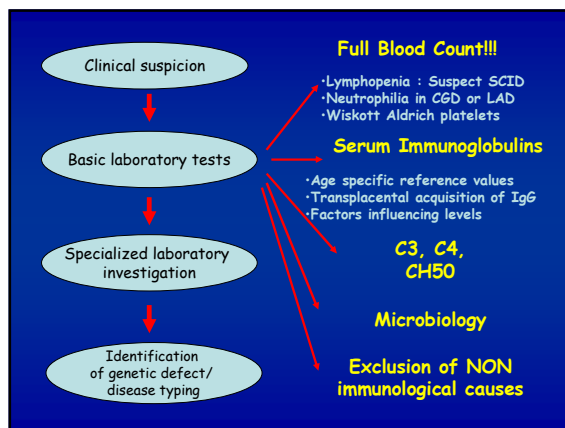
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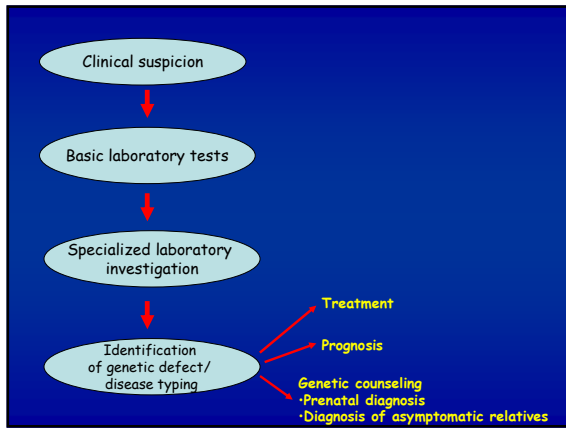
Basic Principles of PID Diagnostic Protocols

1. Rapid diagnosis/exclusion of syndromes in which timely treatment may be life saving, such as:

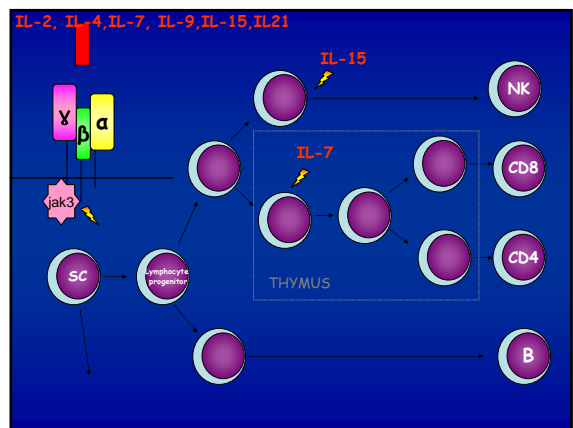
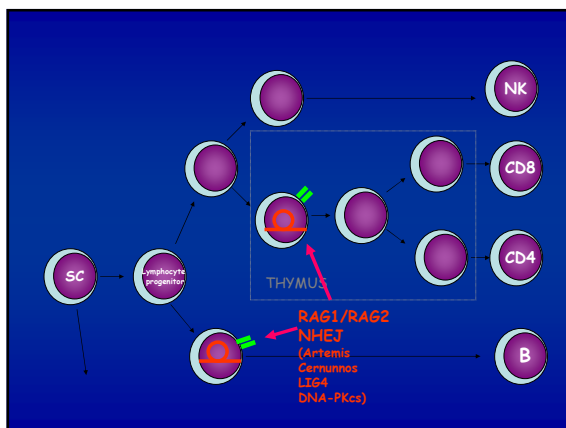
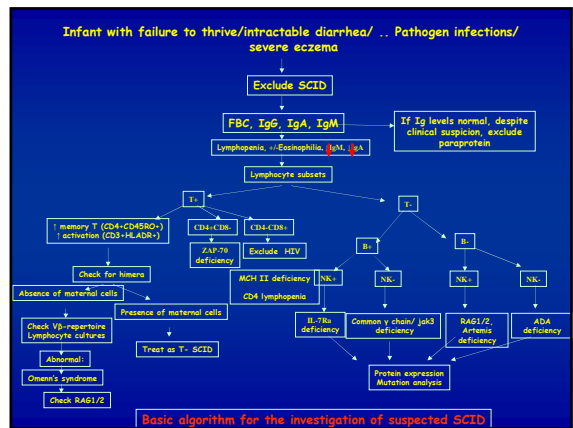
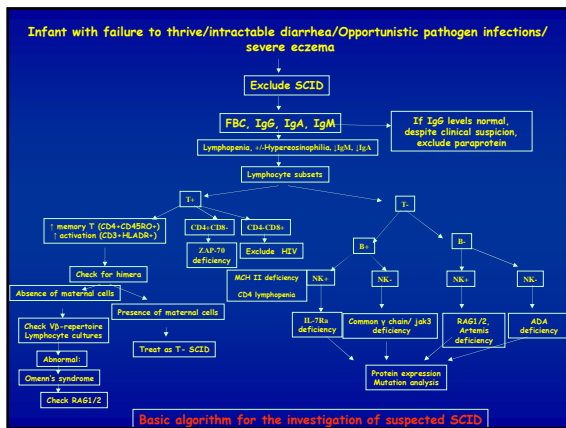
- SCID (Severe Combined Immunodeficiency)
- CGD (Chronic Granulomatous Disease)
- XLA (X-Linked Agammaglobulinemia)

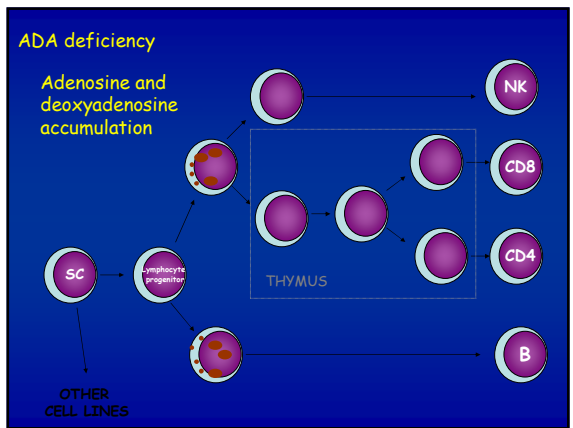
2. Step by Step testing





Severe Combined Immunodeficiency SCID





CAUTION!!!

Presence of T-cells does NOT exclude SCID!!!

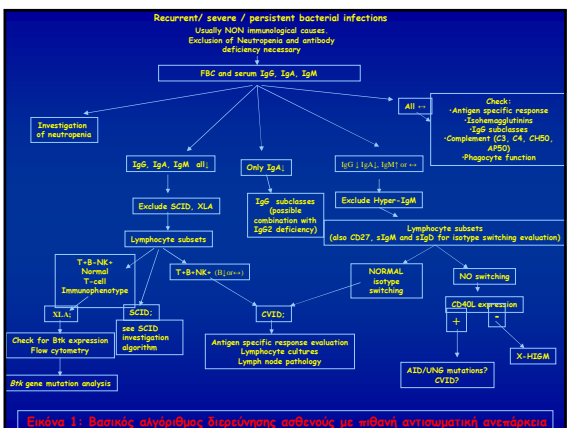
- Maternal T-cells
- Omenn's syndrome

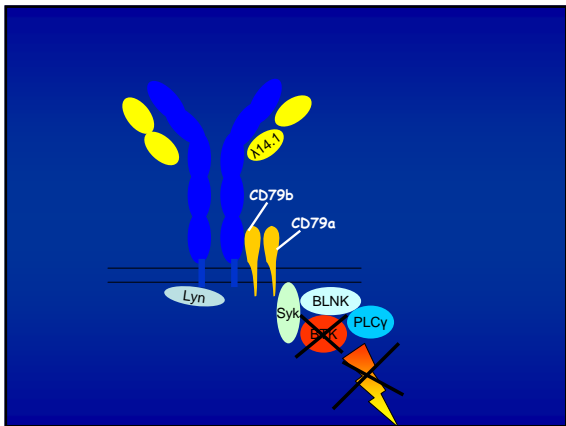
Maternal T-cells → Check for HLA typing

Omenn's syndrome → clonality

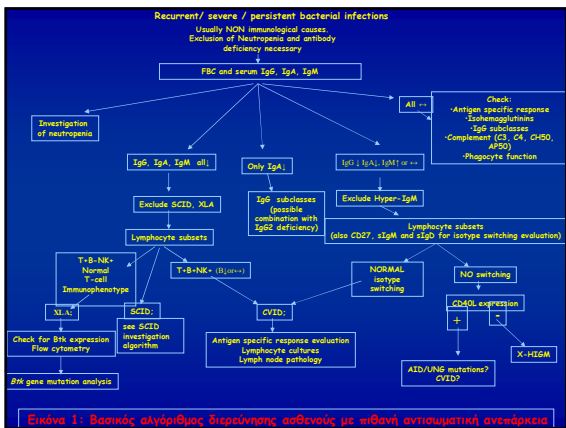
Antibody Deficiencies

Agammaglobulinemia

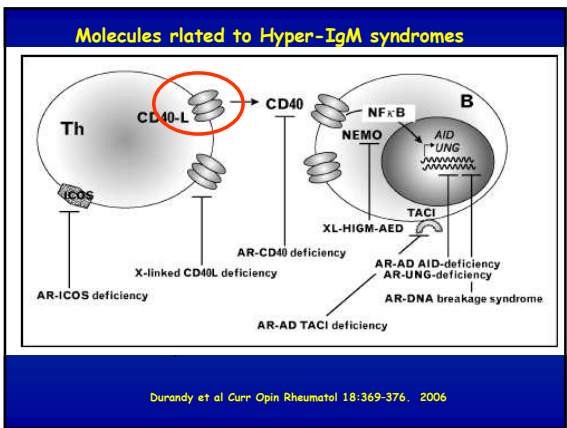




Hyper-IgM syndromes



Εικόνα 1: Βασικός αλγόριθμος διερεύνησης ασθενούς με εθιστή αντισηματική ανεπάρκεια



Durandy et al Curr Opin Rheumatol 18:369-376. 2006

Chronic Granulomatous Disease

