Severe Combined Immunodeficiency

SCID T-B+NK+

An overview made by Ruben de Haan

What is T-B+NK+ SCID?

SCID is a group of auto-immune disorders caused by a mution in genes. T.B.NK. stands for the variant of SCID. For T-B+NK+ this means the patient lacks T-cells but has B-cells and NK-cells.

Basically the patients have no inner protection against bacteria and viruses.

A popular case was the ‘Boy In The Bubble’..

David Vetter aka the ‘Boy in the bubble’,

David lived for 12 years in a sterile bubble to protect him from infections and diseases.

Source: Wiki (wiki/David_Vetter)

Diagnosis:

In the first weeks after birth normally few problems occur, since the protectorcells of the mother are still present in the patient's blood.

±3 months

Then the patient's immune system begins to decline shown by red rash and vulnerablility to infections that do not heal.

You need white bloodcells to protect you.

- White bloodcells = leukocytes
- There are three main kinds:

Type:	T-cells	B-cells	NK-cells
Main function:	Help and kill other cells.	Produce antibodies.	Kill viruses and bacteria.

Problem:

The differences within SCID vary in what is lacking in the patient's blood in terms of leukocytes.

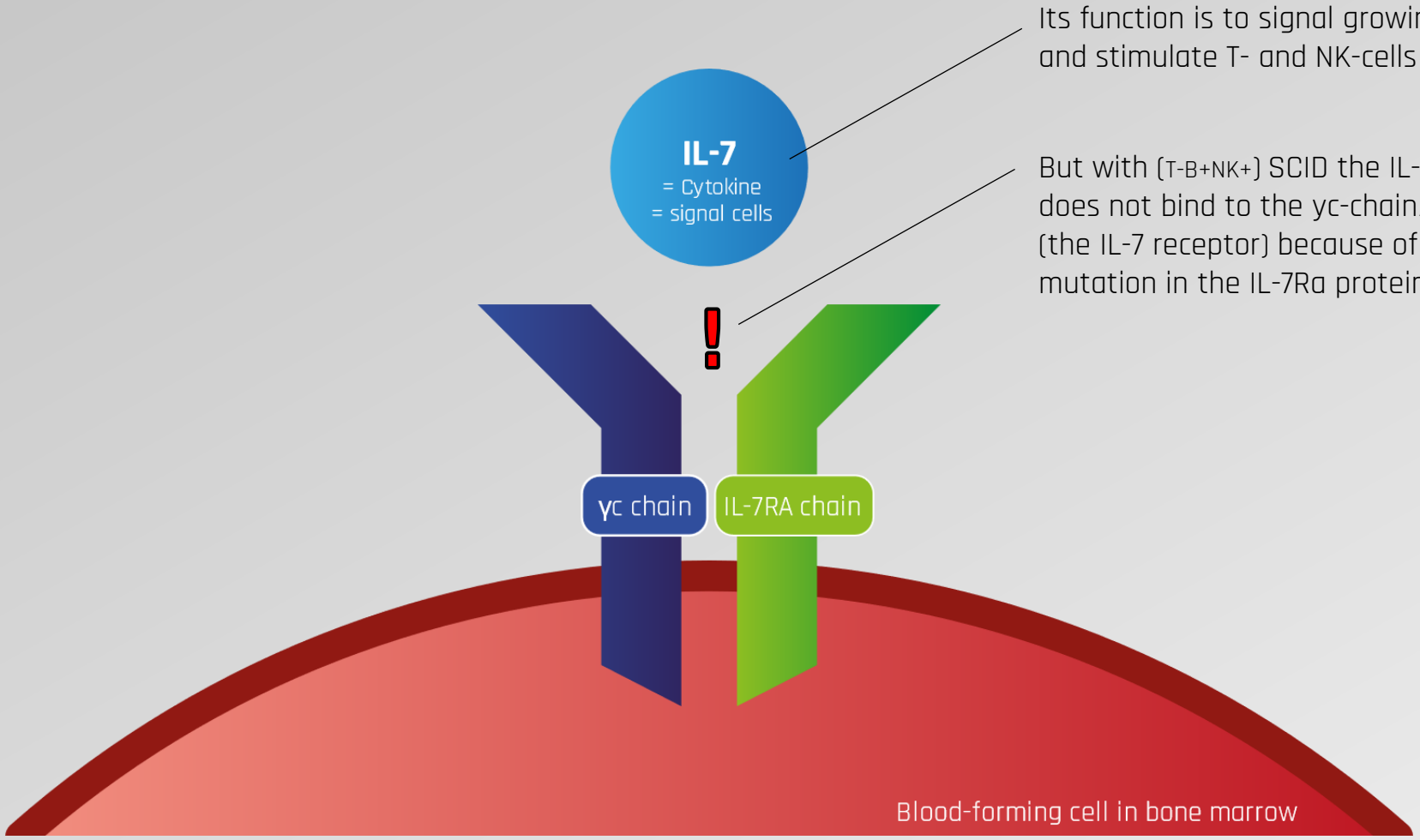
The different types of SCID:

Phenotype	±% of total SCID cases:	Gene Defect:	Pathogenic mechanism:
T-B+NK-	40%	JAK 3, Y-chain	Weakened cytokine signaling
T-B+NK+	10%	IL-7Ra	Weakened cytokine signaling
...	...	...	...

In the case of T-B+NK+ the following occurs:

Source: 'SCID: New and Old Scenarios', Informa Healthcare (www.informa-healthcare.com, 2012)

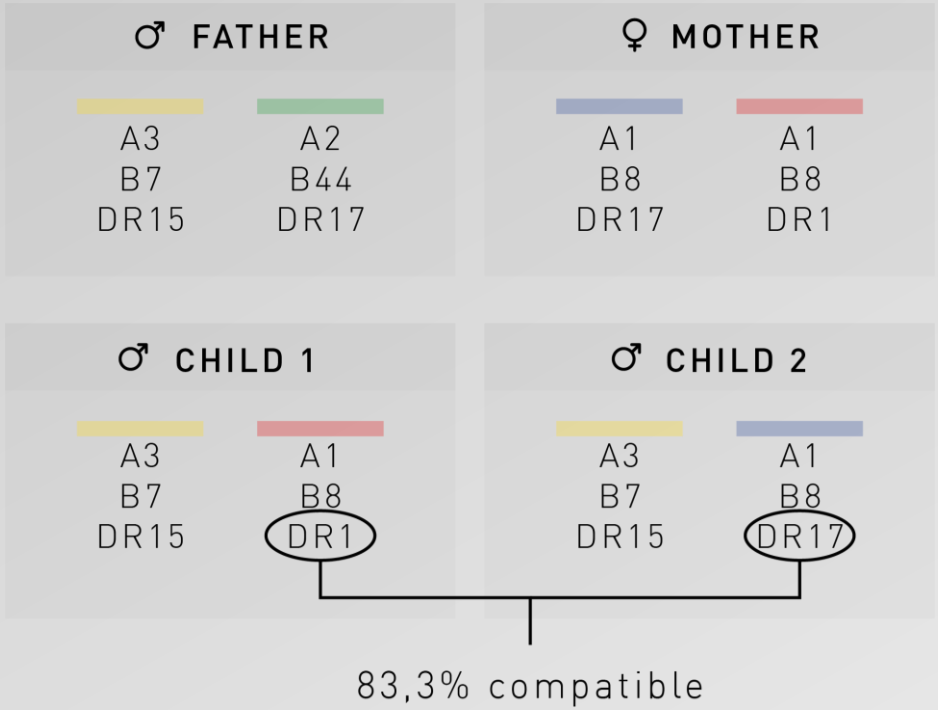
Problem on cellular level:



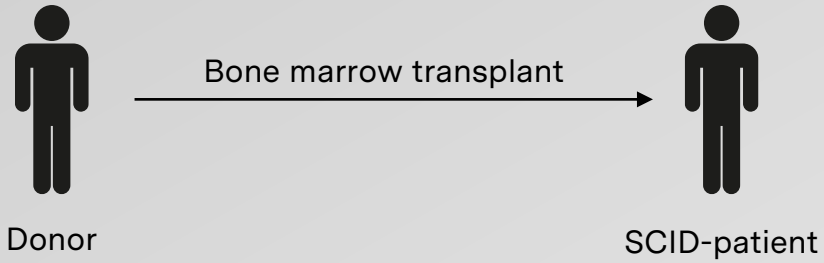
No IL-7 signalling means
no B-cell development & a lowered T-cell and NK-cell development and this means
no working immune system.

Current treatment:

HLA screening:



Current treatment:



Conceptual proposal

Problem:

		Deficient gene:
Cytokine Receptors	sub-unit:	IL2RG
	heterodimer:	IL2 IL4 IL7 IL9 IL15 IL21
	monomer:	IL7RA

Source: ESID patient register (www.esid.org, 2010)