Our Immune System is not Perfect: Some Human Experiments!

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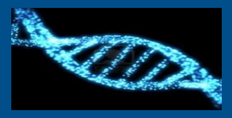
What is a Primary Immunodeficiency Disease (PID)?

Our immune system is composed of many elements that allow our body to combat pathogens. However, some patients lack some of these critical components and are therefore susceptible to life-threatening recurrent infections. Genetic, or primary immunodeficiencies, are inherent in the patient's DNA, and are hereditary (passed from parents to child).



1. DNA Helix, the blueprint of life.

What Basic Methods are used to Treat these PIDs?

- ***Antibiotics*** fight bacterial infections. They either kill bacteria or keep them from reproducing. However, such antibiotics are not a permanent solution for a patient with an unhealthy immune system.

2. Antibiotic Medication

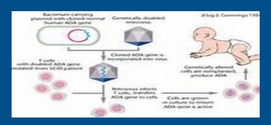
- ***Intravenous Immunoglobulins***, *or* ***IVIG***, inserts antibodies collected from many healthy donors in patients without the ability to produce antibodies. This is a passive immunity as the patients are unable to produce Immunoglobulins on their own.

3. Commercial IVIG.

- ***Hematopoietic stem cell transplantation*** **(HSCT)** is the transplantation of healthy donor stem cells to reconstitute the patient's defective immune system.

4. Cultivation of Stem cells

- ***Gene therapy*** a new technique in which the normal gene is transduced into the patient's defective stem cell and thus repairing the genetic defect.



5. The essential process of gene therapy

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What are some examples of Primary Immunodeficiencies??

Severe Combined Immunodeficiency

SCID is the most severe PID where there is a lack of B and T cells in the patient. This deprives the patient of an acquired immune response. Any contact with the environment is very dangerous because the body has no mechanism to help him defend against common antigens.

6. A patient with SCID l iving in a sterile environment.

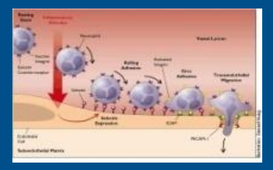
Case Study: Bruton Syndrome(XLA)

XLA is a rare genetic syndrome where B cells, and the cell-producing antibodies, are absent from the body. XLA is X chromosome-linked and therefore almost always limited to males. The syndrome is treated by lifelong periodic infusion of intravenous antibodies.

7. A patient with XLA fighting infection.

Leukocyte Adhesion Disorder (LAD)

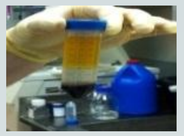
In leukocyte Adhesion Disorder, or LAD, phagocytes are not able to migrate to the site of infection due to their inability to adhere to the blood vessel and move to the tissue. Thus, all phagocytes remain in the vessel and are useless.



8. The mechanisms of neutrophil adhesion .

What are the Basic Methods used to Diagnose PIDs?

This technique is essential for separating the main components of blood: erythrocytes, plasma, and white blood cells. A sample of blood is centrifuged for twenty minutes and the sample is divided into white blood cells, red blood cells, and plasma.



9. Plasma separation

Cell Separation

Methods

Neutrophils are the most abundant type of white blood cell. They engulf foreign particles regardless of their identity. After cell separation, the neutrophils can be seen and counted via microscope.

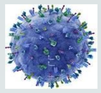
10. Neutrophils as seen through microscope

Neutrophil Detection

B Cell Detection

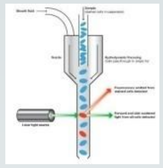
T Cell Detection

B cells produce antibodies that circulate the blood. The FACS machine recognizes B cells by; the CD 19 and 20 markers on their cell surface.



12. B cell

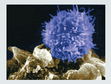
FACS Machine



11. The mechanism of the FACS machine (flow cytometry)

The FACS Machine uses fluorescent markers to identify the different types of white blood cells, particularly B and T cells, in a sample. These are identified by the presence of clusters of differentiation (CD), which are proteins and glycoproteins unique to each cell type.

Helper T Cells (TH) detect pathogens and stimulate the production of antibodies, while cytotoxic T cells (TC) kill infected cells singlehandedly. The amount of T cells is detected by CD3, TH by CD4, and TC by CD 8.



13. T Cell