Severe Combined Immunodeficiency Disorder

What is Severe Combined Immunodeficiency Disorder (SCID)?

It is a rare genetic disorder affecting the immune system, specifically the T and B-lymphocytes. These are white blood cells whose main function is to recognise and fight infection. B cells make antibodies also known as immunoglobulins. Antibodies work by identifying and binding to foreign particles/substances. The spleen and lymphoid organs then remove these antibody bound particles. T cells coordinate immune cells to respond to germs particularly viral and fungal organisms. There are also some T cells, which change into killer cells and attack and destroy the foreign cells.

In Severe Combined Immunodeficiency Disorder both the T and B cells do not work properly, causing the child to have a very poor ability to resist infections. This can lead to serious and often life-threatening infections.

SCID is the result of a defect or mutation in a child’s genes. It may be inherited or due to a new mutation. The most common form is linked to the X chromosome and affects only males, whilst others may be due to an autosomal recessive trait. Yet another type is caused by a deficiency of an enzyme called Adenosine deaminase (ADA). Your Doctor will discuss which type of SCID relates to your child.

Modern research is helping us to know more about SCID and how to treat it. Treatment now helps decrease the risk of serious infection, and can either manage or cure the disorder thus improving the quality of children’s lives.

What are the signs and symptoms?

- Mouth or groin infections (thrush) that are hard to treat
- chronic diarrhoea
- ear infections
- feeding problems, weight loss, failure to thrive
- weakness
• chest infections (pneumonia)
• blood infections (sepsis)
• repeated infections

Germs that do not usually cause problems for healthy children can be overwhelming for a child with SCID due to their poorly working immune system.

What tests will my child have?
• CBP- A simple routine blood test is performed
• Specialised immunity blood tests. These blood tests will measure how well the immune system is working. A child with SCID usually has a very low number of white blood cells, as well as few or no B or T cells. The few cells they may have often do not function properly. Blood tests may also show a very low level of immunoglobulins.
• Chest x-rays and scans
• Sampling of urine, faeces and mucus from the throat.

What treatment is available for SCID and what are the side affects?

Stem Cell Transplant: Children with SCID are often treated with a haematopoietic stem cell transplant. The aim of the transplant is replace the faulty cells with cells from a healthy donor. (Specific information will be given to you if your child is to have a transplant).

Blood transfusions: It is common for a child with SCID to need blood, platelet or plasma (the fluid part of blood) transfusions. The frequency of transfusions may vary and will be discussed with you by your doctor. Modern day screening of donor blood is of a very high standard; hence the risk of infections is very small.

Immunoglobulin therapy: A child with SCID is unable to create enough natural antibodies (blood proteins that kill germs) to fight infection. Treatment called Immunoglobulin therapy helps provide some of the missing antibodies. Immunoglobulin is a solution of purified human antibodies that have been removed from donated blood and is given intravenously (into
the vein). It helps protect against common childhood illnesses, therefore vaccinating with live vaccines such as polio, measles, mumps or rubella is not given whilst on this therapy. Please discuss vaccinations and precautions with your medical team. They will also outline any possible side effects of this treatment.

**Medication:** The use of antibiotics, anti-viral and anti-fungal medication is common in children with SCID to protect against serious infections. Your medical team will provide further education with each medication that your child requires.

**PEG – ADA:** Adenosine deaminase is an enzyme needed to protect immune cells from damage by toxic material. Children who have developed SCID from an ADA deficiency may be given injections containing the missing enzymes to help correct metabolic abnormalities enabling some recovery of immune function and therefore protection against infection.

**Precautions/ useful information**

As a child with SCID is susceptible to infections, it is important to keep your child isolated from people with known infections or contagious illnesses. Caution should be maintained with children or siblings attending day care/school etc. where the risk of catching infections becomes higher. It is essential that hand washing by everyone who comes into contact with your child is carried out.

Vaccinations should be discussed with your doctor including live vaccines for siblings as these may pose a threat to your child who is immunocompromised.

Caring for your child’s skin is vital as it is the body’s first defence mechanism. The child should have a daily wash and the skin be inspected for any broken areas. If you notice any broken areas or signs of infection such as sores that are red, tender, swollen, itchy or blistered it is important to let the medical team know. General signs of infection may include a temperature above 38.5, tiredness, aches and pains or a headache.

A balanced nutritional diet is important for your child. In many instances, a child with SCID cannot absorb food normally, which can lead to poor nutrition. A dietician within the WCH can assist you to provide adequate nutrition.
References and Further information:

The Internet is a source of vast amounts of information about SCID, its treatment and the function of the immune system. Please be mindful that some information may not be correct or relate to your own child – as anyone can display information on the World Wide Web. The best way to find appropriate information is through reputable sites such as:

The SCID homepage = http://www.scid.net/ and follow their prompts.

www.gosh.nhs.uk/factsheets
www.primaryimmune.org/pubabook_pats/e_cwo5.pdf
www.niaid.nih.gov/final/immun/immun.htm

Please refer to section 7 “Sources of More Information” or check the parent’s library in McGuiness & McDermott children’s clinic for further information.