X-SCID

X-linked severe combined immunodeficiency (X-SCID)

Additional information for families

www.immunodeficiencyuk.org
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0800 987 8986

Great Ormond Street Hospital for Children NHS Foundation Trust

THE great north CHILDREN'S HOSPITAL

IMMUNO DEFICIENCY UK

Supporting families affected by primary and secondary immunodeficiency

About this leaflet

This leaflet has been produced jointly between Immunodeficiency UK, Great Ormond Street Hospital (GOSH) and the Great North Children's Hospital. It describes the X-linked specific form of severe combined immunodeficiency (SCID) and **should be read in conjunction with the general overview leaflet on SCID available on our website at www.immunodeficiencyuk.org.**

The information has been reviewed by the Immunodeficiency UK Patient Representative Panel and by families affected by X-SCID and endorsed by the Immunodeficiency UK Medical Panel but should not replace advice from a clinical immunologist or a geneticist.

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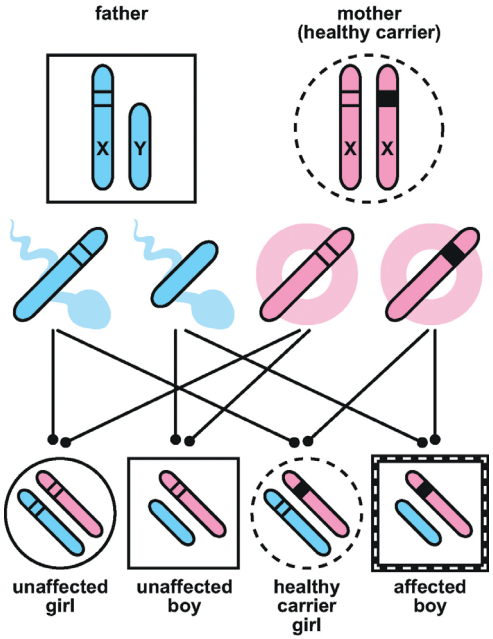
What is X-SCID?

X-SCID is a form of severe combined immunodeficiency (SCID) that is inherited as an X-linked disorder. This means that only males can be affected and that females can be carriers. There may be a history of SCID in other males in the family (maternal uncles, male cousins on the mother's side), but often it occurs as a 'new mutation' that has developed 'out of the blue' or sporadically, in which case there is no previous family history. More information about X-linked inheritance can be found in the leaflet *Genetic aspects of primary immunodeficiency*, available on our website at **www.immunodeficiencyuk.org**.



Mechanism of X-linked inheritance

What causes it?

X-SCID is caused by a mistake (mutation) in a gene on the X chromosome. The affected gene codes for a protein known as the 'common gamma chain' (γ_c). γ_c is very important for normal development and functioning of a number of cells that are vital for a healthy immune system. These include T cells, natural killer (NK) cells and B cells. In X-SCID, γ_c is either absent or not working, and this results in a severely abnormal immune system.

What are the signs and symptoms?

The signs and symptoms in babies with X-SCID are the same as in all forms of SCID, as described in our accompanying leaflet available on our website at **www.immunodeficiencyuk.org**.

How is it diagnosed?

Initial investigations are the same as for all forms of SCID, as described in our accompanying leaflet.

There are a number of features that might suggest a diagnosis of X-SCID. A detailed family history is important. If the baby is a boy and there is a history of affected boys in the family (including unexplained infant deaths), X-SCID may be likely. In addition, the basic immunology tests looking at the presence of T, NK and B cells may show a pattern suggestive of X-SCID. The pattern doctors are looking for is 'T- B+ NK-', meaning that T cells and NK cells are absent or very low in number, while B cells are present in normal numbers. If this pattern is confirmed, particularly when the family history is suggestive, X-SCID is likely. The next step is to perform specialised tests to look for the γ_c protein and to carry out genetic testing to look for a mutation in the γ_c gene. Results of these tests are usually available within a few days.

How is it treated?

The basic approach to looking after an infant with X-SCID is the same as for all forms of SCID, as described in our accompanying leaflet available on our website at **www.immunodeficiencyuk.org**.

In the UK there are two specialist centres in the country that provide definitive treatment for children with SCID – Great Ormond Street Hospital (GOSH) in London, and the Great North Children's Hospital (GNCH) in Newcastle. Definitive treatment begins when the child is referred to one of these centres. Initial stabilisation and treatment under advice may begin in a referring specialist children's hospital or treatment centre.

When the time comes to discuss definitive (curative) treatment, there may be a choice between haematopoietic stem cell transplant (HSCT) and gene therapy. X-SCID is one of the small number of conditions for which clinical trials in gene therapy have been successful. The decision regarding which treatment is best will depend on the availability of well-matched donors for HSCT. This will require extensive discussion with the transplant and gene therapy teams.

What does this mean for the future?

Genetic counselling is important not only for the immediate family but also for the extended family on the mother's side, because other relatives might be carrying the faulty gene. For females identified as carriers, prenatal diagnosis is possible for future pregnancies. Referral to local genetic counselling services can be arranged, and in some situations a joint counselling appointment with a genetics specialist and an immunologist can be helpful. More information about the genetic aspects of primary immunodeficiency is available on our website at **www.immunodeficiencyuk.org**.

Is there a support group?

Immunodeficiency UK is the main support organisation in the UK for anyone affected by a primary or secondary immunodeficiency disease. Call our helpline on **0800 987 8986** or visit our website at **www.immunodeficiencyuk.org**.

It can be helpful to meet another family who has a child with SCID and who has undergone HSCT or gene therapy. Speak to your immunology team, who may be able to arrange a meeting with a suitable family.

The leaflet *How to become a bone marrow donor* can be obtained from the Anthony Nolan Bone Marrow Trust by ringing **0303 3030303** or visiting their website at **www.anthonynolan.org**.

Notes

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About Immunodeficiency UK

Immunodeficiency UK is a national organisation supporting individuals and families affected by primary and secondary immunodeficiency.

We are the UK national member of IPOPI, an association of national patient organisations dedicated to improving awareness, access to early diagnosis and optimal treatments for PID patients worldwide.

Our website has useful information on a range of conditions and topics, and explains the work we do to ensure the voice of patients with primary and secondary immunodeficiency is heard. If we can be of any help, please email us or call on the number above, where you can leave a message.

Support us by becoming a member of Immunodeficiency UK. It's free and easy to do via our website. Members get monthly bulletins.

Immunodeficiency UK is reliant on voluntary donations. To make a donation, please go to **www.immunodeficiencyuk.org/donate**

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