

## **This all seems very complicated, so what practical advice can you give?**

All boys with XLI should be examined by a doctor to see if:

- both testicles are present and descended
- penile size is normal
- growth or height is normal (as expected from parental height)
- mental development is normal
- puberty is normal

If any of the above are abnormal further investigation by specialist hospital doctors may be needed. All the above problems can be treated.

## **2) If you are a carrier for XLI. (i.e. have a child with XLI)**

If you are a mother of a child with XLI you may develop a problem with future pregnancies. As a female carrier of XLI you have rather low levels of steroid sulphatase but you have enough to give you normal skin. However, your placenta also makes steroid sulphatase and this doesn't always make enough to function normally. The only problem that can sometimes arise is that labour (childbirth) may be very prolonged. This in turn can damage the unborn baby if the labour is not being adequately monitored.

## **So what practical advice can you give?**

When you know you are pregnant book in early at your local hospital pre-natal clinic. Tell your obstetrician (baby doctor) that you have steroid sulphatase deficiency. They can take appropriate precautions at the time of delivery to prevent any birth complications. Induction of the birth or Caesarian section are occasionally needed. Do not have a home delivery. Your childbirth will need to be closely monitored to prevent unnecessary damage to your child.



**Ichthyosis Support Group**  
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## **X-Linked Ichthyosis**

### **What is it?**

X-linked ichthyosis (XLI) is a condition due to an inherited deficiency of an enzyme called steroid sulphatase. This enzyme is found throughout the body but seems most important in the skin. When there is too little enzyme the skin can't "shed" normally so it appears dry and scaly. All of the skin can be affected but the face, scalp, palms and soles are relatively spared. The skin scales can look rather brown or pigmented at times and this can lead to teasing at school as people misinterpret this as being an "unwashed appearance". Of course it has nothing to do with personal hygiene. The abnormal gene that causes XLI is carried on one of the sex chromosomes called the X-chromosome; hence the name of the disorder. XLI only occurs in boys (who have 1 X-chromosome). Girls (who have 2 X-chromosomes) can be carriers but their skin is usually normal (see separate sheet on inheritance of ichthyosis). The severity of scaling in XLI varies enormously even within the same family. It is most marked in the winter (especially dry cold weather) and some individuals show relatively normal skin in the summer. The scaling is rarely severe enough to cause problems with decreased sweating or pulled down eyelids ("ectropion") that occur in the more severe ichthyoses. Surprisingly the deficiency of steroid sulphatase elsewhere in the body only rarely causes other medical problems (see later).

### **Is it catching?**

No this is not catching or contagious.

### **When will I know if my child is affected with XLI?**

Normally the skin scaling is apparent at or shortly after birth. A few individuals do not develop scaling for many months but nearly every affected individual will show signs of XLI by 1 year of age.

### **Will I grow out of it?**

In one sense the answer is “no” as the enzyme deficiency lasts lifelong. However more than 80% of people with XLI say that their skin improves with age, especially by adult life. Many adults with XLI have relatively normal looking skin in the summer months. The mechanism for this improvement with age is not understood but presumably the skin must adapt in some way to compensate for the lack of sulphatase.

### **How common is it?**

It is relatively common compared to other types of ichthyosis and is estimated to occur in 1/2000–1/4000 male births. It is likely that many mild cases remain undiagnosed.

### **How is it treated? (see sheet on topical therapies for more details)**

Most individuals with XLI are treated with topical creams and bath oils. These include moisturisers and creams containing urea or lactic acid. It is important to try a number of these products to find which is most suitable for you. It may be that you will need to use a more greasy product (an ointment rather than a cream) in the winter when the condition is usually worse.

Occasionally in very severe cases of XLI a drug taken by mouth (called acitretin or neotigason) may be considered but this can have some side effects so this needs to be discussed with your doctor.

### **Can it be cured?**

No not at the moment. However if “gene therapy” (i.e. replacing defective genes) ever becomes a reality in medicine, XLI would be an obvious condition to treat as there is only one gene involved and the skin is easily accessible. However as XLI is not life-threatening it remains to be seen whether such a potentially expensive type of therapy would ever be used.

### **Will my children inherit it? (see sheet on inheritance)**

If you are a female carrier for XLI (i.e. have an affected son but are unaffected yourself) you will have a 1 in 2 chance of having an affected boy and a 1 in 2 chance of having an unaffected girl who is

a carrier. If you have XLI your children will be unaffected. However your sons will be entirely normal and all your daughters will be carriers for XLI. Therefore there is a risk of XLI for your male grandchildren.

### **Are there any associated medical problems?**

- 1) If you have XLI
- 2) If you are a carrier for XLI

#### **1) If you have XLI**

#### **We must stress that most boys with XLI have no other associated medical problems.**

Rarely there are other medical problems although the reasons are rather complicated but an understanding of the genetics may help to explain.

XLI is an unusual genetic condition in that rather than have a small mistake (mutation) in the steroid sulphatase gene, it is more normal to be missing the whole gene. When this gene is deleted it may also “knock out” the genes next door. On one side there are genes for mild mental retardation and short stature. Therefore if these genes are missing an affected boy will have learning difficulties and be short in height as well as having scaly skin. On the other side is a gene (the “Kallmann’s gene”) which controls normal puberty. If this is missing an affected boy will not go through puberty unless hormone replacement is given. There may also be other subtle problems with hand movements, the ability to smell and the kidneys (so-called Kallmann’s syndrome). Other boys with XLI have trouble with normal puberty or failure of one testicle to descend even though they don’t have Kallmann’s syndrome. The reason is not known but it may be due to some problem with steroid sulphatase enzyme levels.

Unfortunately most doctors are not aware of the above associated medical problems so you may have to fight to get the appropriate help.