



Harlequin Ichthyosis

This is an extremely rare and severe inherited (genetic) ichthyosis. There are approximately five such children born in the UK each year and some may be stillborn. The name comes from the appearance of the skin at birth.

What does it look like?

Affected babies are born prematurely and small for dates. The skin is covered in tight, thick, plates of hard scale and resembles armour plating or the harlequin suit of a jester. It splits at several areas causing deep cracks. The face looks stretched with turned out lips and eyelids, and the ears, hands and feet may be hidden in the scale. The eyes may be invisible because they are temporarily covered by the swollen eyelids. The baby's movement and ability to suck is affected and they suffer all the problems of leaky skin. They become dehydrated and have poor temperature control, difficulty feeding and sometimes breathing and are also at risk of serious infection.

What happens next?

The baby will be nursed in an incubator and have intensive care to cope with all the expected problems. Skin creams and perhaps retinoid medicine will help the scale plates to shed in a few weeks. Then the skin will appear red, tight and scaly all over and this is usually an ongoing problem. The eyes reappear and may need to be protected with bandages or a small operation as the eyelids do not cover them fully. Special feeding and other treatments are often needed and things may not always go well. Children who survive are affected with severe inflamed ichthyosis and may be small and scarred in places. They will need ongoing intensive skin treatment with creams and courses of retinoid medicine and many require physiotherapy, counselling and extra tuition.



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What is the cause and can it be prevented?

Harlequin ichthyosis comes out of the blue. The parents of an affected baby are carriers and will have a one in four risk of any future baby of theirs being similarly affected. This is another genetic disease due to a single important skin gene being faulty or mutated. In this case, the mutation is in ABCA12, a gene thought to be involved in transport of lipids (fats) into the spaces between the cells in the skin's uppermost layer. These lipids act as a protective barrier against bacteria and infection.

Harlequin ichthyosis cannot be prevented but it is possible to diagnose or recognise it early in pregnancy by removing some of the amniotic fluid surrounding the developing foetus to identify if there is a mutation in ABCA12. This allows the family to decide if they wish to continue or terminate the pregnancy. It is also feasible that preimplantation diagnosis may be available in the near future. Preimplantation testing is a relatively new way to look even earlier into the genes of the developing embryo. In preimplantation diagnosis a normally fertile couple will choose to undergo in-vitro fertilization and then test each of the embryos for a genetic disease, in this case harlequin ichthyosis, before they are transferred to the mother's womb. Preimplantation diagnosis is difficult, costly and not widely available, but is still an exciting possibility for parents who have already had a child with harlequin ichthyosis. The finding of a gene for harlequin ichthyosis may also allow development in the future of a specific treatment to replace the missing protein in the upper layers of the skin in this disease.

There are families with one or more children with Harlequin ichthyosis who live happy and relatively normal lives.



Ichthyosis Support Group
September 2005

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