HITS (UK) MEDICAL ARTICLE NUMBER ONE

Information for Parents

Ito's Disease

Dr Ito first wrote in a Japanese medical journal in 1952 about some patients he had seen who had a pattern of skin markings in a V shape over the spine and in lines down the arms and legs. Over the years other doctors have written in medical journals describing one or two patients with the same skin markings. Some of the patients have had problems with development. Until recently no one knew the cause of Ito's disease but we have done research in Manchester and our results have been confirmed by other studies in the USA.

What causes Ito's disease?

The body is made up of millions of cells usually each one has the same genetic instructions as each other (chromosomes and genes). In Ito's disease we know that there are two groups of cells, one with one set of genetic instructions and another with a slightly different set of instructions. It is the upset in "balance" between the two sets which causes the problem.

Do all children with Ito's disease have the same variation in genetic instructions?

No, some children have an extra chromosome or chromosomes in one set of cells whereas other children have a variation in one of the smaller units called genes, which cannot be seen down the microscope.

How does Ito's disease come about?

In the type with the extra chromosomes usually the egg and the sperm are normal when they join together. The fertilised egg then divides and it is at this division or one of the following divisions that the chromosomes do not divide equally giving rise to a set of cells with an extra chromosome.

In the type with the gene variation the egg and sperm contain normal gene instructions and when the fertilised egg divides a gene fault occurs in one of the cells and thereafter all the cells resulting from that cell contain the faulty gene.

The above problems just happen by chance and are not caused by anything the parents have done and are not usually inherited.

Are all children with Ito's disease the same?

No. How a child is affected depends partly on the particular chromosome or gene fault and partly on how many cells contain the fault.

What sort of problems do children with Ito's disease have?

These can be divided into two types : Physical and Developmental. Not all children have all of the problems.

Physical Problems

These include problems obvious at birth such as unusual shape of joints, extra fingers, etc. Many children with Ito's disease have more growth on one side of the body than the other making them a little asymmetrical.

The skin patterns are usually first noted when a child is a few months old.

Developmental Problems

Some children with Ito's disease make very good progress and others are slow with their development. There is no way other than careful follow-up to predict how a particular child will develop. Even those children with problems with development do continue to grow and make progress although slower than other children. Extra help in nursery and school may be necessary.

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Will my next baby have Ito's disease?

No, our research has shown that in almost every case of Ito's disease it is not inherited and only affects one child in a family. This is because the egg and sperm are usually absolutely normal and the fault occurs as a one off problem after the cells have started to divide in the developing baby.

How many people are affected and is research continuing into Ito's Disease?

We believe it is a rare condition and even specialists in genetics have each only seen a few patients. However, our research is continuing to learn more about Ito's disease and we will inform our families of any advances.

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Further medical articles are available from Brenda Rugeley (*note these will incur a small charge to cover the costs of printing and postage) – please e-mail Brenda on brenda.rugeley@tesco.net@internet

Thank you

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