# Incontinentia Pigmenti (IP)
# (Bloch-Sulzberger syndrome)

by

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## How common is IP?

Incontinentia Pigmenti is a rare condition, affecting girls. Only in exceptional circumstances does it affect boys.

## What are the features of IP?

IP affects many parts of the body. It can affect different people to different degrees.

### Skin features

There are typically four stages that occur one after the other, though they may overlap. No specific treatment is needed for the skin changes.

Stage 1

There is redness of the skin, then blistering, starting from the first few weeks of life. The blisters do not affect the face but occur in lines along the limbs and round the body. Blistering usually stops by four months of age, though blisters may recur at times when the child has a temperature. During this stage, the blisters should be kept clean and dry.

Stage 2

As the blisters heal, warty areas occur on the skin of the hands and feet. In most cases these clear by six months of age.

Stage 3

This is the stage that gives the condition its name. There are streaks and whorls of pigment along the limbs and round the body. These darken initially, then fade, usually by the age of 16 years.

Stage 4

In adults there are pale, hairless streaks, best seen with an ultra violet light on the backs of the calves.

### Nails

About 40% of people with IP have fragile or ridged nails. Some have non-cancerous lumps under the nails that can be painful and can be removed surgically.

### Hair

Some girls with IP have thin hair and a few may have bald patches during the blistering stage. Later, half the people with IP have course, dull hair. Hair colour is normal.

### Eyes

One third of girls with IP have a squint. They should be checked to see if they need glasses in order to prevent more serious visual problems.

More than 90% of people with IP have normal vision but some have a problem with the blood vessels in the back of the eye (retina). If present, this usually only causes a problem with one eye. Girls should have their eyes checked each month for the first few months. In some cases specialist treatment of the eye may be required.

### Teeth

Over 80% of people with IP have late eruption of the teeth or fewer teeth than normal. Sometimes the teeth are a conical shape.

### Breast

A small number of women may have some asymmetry in the size and shape of their breasts.

### Development

Early studies showed that girls with IP often had problems with their development but more recent and more accurate studies have shown this not to be the case. Less than 10% of girls with IP have developmental or learning problems. Those who do have problems with development may have fits in the newborn period.

## Genetics of IP

IP is caused by a change in a gene called NEMO. This gene is on the X chromosome. We have a total of 46 chromosomes in each cell of our body. Most of these are the same in both sexes but females have two X chromosomes and males have an X and a Y chromosome. If a male fetus had an altered NEMO gene it would almost certainly miscarry. Females can tolerate having one altered NEMO gene on one X chromosome as the other, normal gene on the other X chromosome, dilutes the effect of the altered gene.

In some cases the altered NEMO gene is passed from mother to daughter. In other cases the NEMO gene is normal in both parents but a change occurs in the gene when it is passed to the egg or sperm that made the girl.

## Genetic testing

Since the recent discovery of the NEMO gene, it has been possible to test the gene. This may be done to confirm the diagnosis and to see whether the mother is affected. This may be important to establish whether IP could occur again in other pregnancies. Testing in pregnancy may be offered.

## For more information about the genetics of IP

You can ask to be referred to your nearest Department of Clinical Genetics.

This article was first published in our newsletter (Volume 2 Issue 4 - July 2002).