Incontinentia Pigmenti

Incontinentia pigmenti (IP) is a rare, genetic disorder characterized by unusual patterns of discolored skin. Genetic disorders are caused when there are problems with the arrangement of the "genes" or protein codes inside of the cells of our body. The disorder is caused by excessive deposits of melanin (normal skin pigment). Because of the genetic cause of this problem, if a woman has one daughter with this problem, there is a 50% chance that other female infants will also have the problem. Male fetuses are usually miscarried.

SYMPTOMS

IP is divided into 4 stages. These stages frequently overlap or appear together.

- During the first stage, which begins between birth and 6 months of age, areas of skin get red. The skin may also show spiral lines of small fluid-filled blisters.
- The second stage develops slowly with rough, warty skin growths. These growths appear on the arms or legs. Sometimes, they develop on the head or trunk. These growths, which are often arranged in the same spiral or linear pattern as in the first stage, usually go away during infancy or early childhood.
- The third stage begins between 3 months and 2 years of age. During this stage, the skin gets discolored in unusual patterns.
- In the fourth stage, there is loss of skin pigment and/or a shrinking and thinning of the affected skin.

Other problems include:

- Diminished vision (Eye problems occur in about 30% of cases of incontinentia pigmenti, including:
  - Abnormalities of the blood vessel of the retina or the inner lining at the back of the eye
  - Cataracts
  - Crossed eye (lazy eye) in girls.
- Hair loss with scarring.
- Dental problems (Delayed tooth growth or decay, missing or malformed teeth).

Other rare problems include:

- Seizures (convulsions).
- Muscle spasms.
- Spastic paralysis may occur.
- Skeletal problems.
- Wooly or kinky hair.

TREATMENT

Genetic counseling is a good idea to understand the risks and issues.

The skin problems of IP usually disappear by adolescence or adulthood without treatment. Diminished vision may be treated with corrective lenses or in severe cases, surgery. Children with IP should have a pediatric ophthalmologic evaluation as soon as possible after birth and regularly thereafter in the first few years of life. Dental problems may be treated by a specialist. Nervous system symptoms such as seizures and muscle spasms, may be treated with medicine and/or medical devices as advised by a neurologist.

WHAT WILL HAPPEN

Although the skin abnormalities usually regress, and sometimes disappear completely, there may be residual neurological difficulties.

RESEARCH BEING DONE

Research is continually being done to learn more about this problem. Research on IP is aimed at increasing knowledge and understanding of the disorder, and finding ways to prevent, treat, and cure it. One of the key

centers for this research is The National Institute of Neurological Disorders and Stroke.

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