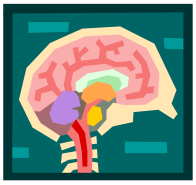

Disability Awareness Begins With You: Incontinentia Pigmenti

What is Incontinentia Pigmenti?
Incontinentia pigmenti (IP) is one of a group of gene-linked diseases known as neurocutaneous disorders. These disorders cause characteristic patterns of discolored skin and also involve the brain, eyes, nails, and hair. In most cases, IP is caused by mutations in a gene called NEMO (NF-kappaB essential modulator). Males are more severely affected than females. Discolored skin is caused by excessive deposits of melanin (normal skin pigment). Most newborns with IP will develop discolored skin within the first two weeks. The pigmentation involves the trunk and extremities, is slate-grey, blue or brown, and is distributed in irregular marbled or wavy lines. The discoloration fades with age. Neurological problems include cerebral atrophy, the formation of small cavities in the central white matter of the brain, and the loss of neurons in the cerebellar cortex. About 20% of children with IP will have slow motor development, muscle weakness in one or both sides of the body, mental retardation, and seizures. They are also likely to have visual problems, including crossed eyes, cataracts, and severe visual loss. Dental problems are also common, including missing or peg-shaped teeth. A related disorder, incontinentia pigmenti achromians, features skin patterns of light, unpigmented swirls and streaks that are the reverse of IP. Associated neurological problems are similar.

Is there any treatment?
The skin abnormalities of IP usually disappear by adolescence or adulthood without treatment. Diminished vision may be treated with corrective lenses, medication, or, in severe cases, surgery. A specialist may treat dental problems. Neurological symptoms such as seizures, muscle spasms, or mild paralysis may be controlled with medication and/or medical devices and with the advice of a neurologist.

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

What research is being done?
Researchers have begun to use genetic linkage studies to map the location of genes associated with the neurocutaneous disorders. Research supported by the NINDS includes studies to understand how the brain and nervous system normally develop and function and how they are affected by genetic mutations. These studies contribute to a greater understanding of gene-linked disorders such as IP, and have the potential to open promising new avenues of treatment.

National Organization for Rare Disorders (NORD)
P.O. Box 1968, 55 Kenosia Avenue
Danbury, CT 06813-1968
orphan@rarediseases.org
http://www.rarediseases.org
Tel: 203-744-0100, 800-999-NORD (6673)
Fax: 203-798-2291

National Eye Institute (NEI)
National Institutes of Health, DHHS
31 Center Drive, Rm. 6A32 MSC 2510
Bethesda, MD 20892-2510
2020@nei.nih.gov, http://www.nei.nih.gov
Tel: 301-496-5248

National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS)
National Institutes of Health, DHHS
31 Center Dr., Rm. 4C02 MSC 2350
Bethesda, MD 20892-2350
NIAMSinfo@mail.nih.gov
http://www.niams.nih.gov
Tel: 301-496-8190, 877-22-NIAMS (226-4267)

From: http://www.ninds.nih.gov/disorders/incontinentia_pigmenti/incontinentia_pigmenti.htm