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The National Ichthyosis Foundation

ICHTHYOSIS FOCUS

THE NATIONAL ICHTHYOSIS FOUNDATION
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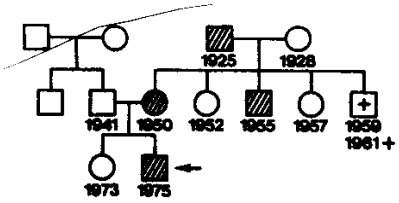
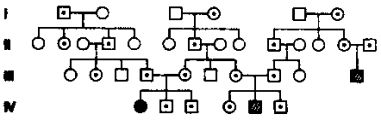
FROM A MEDICAL POINT OF VIEW



DOMINANT DISORDER
Epidermolytic Hyperkeratosis (or) ichthyosis vulgaris

In the above diagram a six year old boy with ichthyosis born in 1975 has a 31 year old mother (age 25 when he was born). His father was 34 when the boy was born. The grandfather, who has ichthyosis, was born in 1925. His affected daughter was born when he was 25. The patient also had an uncle who died at age 2 in 1961. This is the form of inheritance found in dominant disorders. Circles represent females, squares represent males. Birthdates of children and parents are useful for determining whether parental age influences the appearance of the disease.

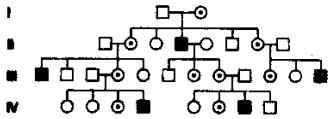
In recessive disorders, both parents must carry the abnormal gene which is usually unexpressed as a recognized disorder. If each parent transmits the abnormal rather than the normal gene to the affected child, the disease will be expressed in the child. Children who receive only one gene for the disorder from either the mother or father will carry that single abnormal gene — but will not have signs of the disorder because the expression of a recessive disorder requires the presence of two abnormal genes. Lamellar ichthyosis is a recessive disorder.



RECESSIVE DISORDER
(Lamellar Ichthyosis)

In the above pedigree, there are two children affected with lamellar ichthyosis in the fourth generation and an affected man in the third generation. The symbols with dots represent carrier state. These "carriers" are individuals who carry the gene for lamellar ichthyosis but do not have ichthyosis. In this theoretical pedigree there is an unusually high incidence of the gene for lamellar ichthyosis.

X-linked recessive disorders such as X-linked ichthyosis are expressed completely only in males. The gene is carried by their mothers who may have minor problems but not the full expression of X-linked ichthyosis. The inheritance is called X-linked because the gene for the disorder is on the X-chromosome. Females have two X-chromosomes, males have one. The mother of an affected boy has two X-chromosomes, one carries the abnormal gene — the other carries a normal gene. Half of the eggs of the mother will contain the abnormal gene, half the normal gene. If the egg containing the normal X-chromosome is fertilized by a sperm containing a Y-chromosome the boy will not have X-linked ichthyosis. If the egg carrying the gene for ichthyosis is fertilized by a sperm containing a Y-chromosome the son will be affected. The affected son's only X-chromosome is abnormal and if he fathers a female child, she will receive this X-chromosome containing the abnormal gene. Since the X-chromosome carries the abnormal gene, all of his daughters will be carriers of the ichthyosis gene. His sons will be normal and will not be carriers since the father has contributed a Y-chromosome to them — not an abnormal X-chromosome.



X-LINKED RECESSIVE DISORDER
(X-linked ichthyosis)

The three pedigrees illustrated above are idealized to demonstrate the three major patterns of inheritance. The ability to make helpful predictions for family planning is based on the study of pedigrees of many families of various sizes with inherited disorders. More data are needed on the transmission of the ichthyoses in order to ascertain the risks for subsequent children in the same generation as the affected individual as well as the risks for children in future generations. That kind of information can be obtained by you. Those of you who would like to help can consult your relatives and construct family pedigrees. Send the information and the basis of the clinical diagnosis to the Newsletter. The information will be used but you will not be identified by name, age, hometown or initials.

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