FRIED AND WITKOP SYNDROMES

There are at least two types of Ectodermal Dysplasia (ED) that affect only the teeth and nails. The features of the two types are similar, consequently they have each been called Tooth and Nail ED at various times. Subtle differences in the features and different patterns of inheritance can be used however, to distinguish between the two types.

Fried Syndrome

In the mid-70s an investigator named Fried (rhymes with feed) described a type of ED whose features include sparse, fine hair, hypodontia (congenital absence of several teeth), thin fingernails and small flat toenails. Sweat function, hearing, vision, intelligence and life span are normal. Fried syndrome is inherited as an autosomal recessive, meaning that it is caused when someone has two copies of the responsible gene, one from each parent. The parents "carry" the gene, but are not affected. Females are affected by Fried syndrome as often and as severely as males.

Witkop Syndrome

In the mid-60s another investigator, named Witkop *(rhymes with stop)*, described a type of ED whose features include fine hair that may be somewhat sparse, hypodontia (congenital absence of several teeth) and small, spoon-shaped finger and toe nails. Sweat function, hearing, vision, intelligence and life span are normal. Witkop syndrome is inherited as an autosomal dominant, meaning that it is caused by a single copy of the responsible gene. The copy of the gene is usually inherited from one or the other parent. The parent with the gene is affected. Females are affected by Witkop syndrome as often and as severely as males.

The two syndromes then are quite similar and the normal variation in the way things show up makes diagnosis difficult in any given person. The best way to differentiate between them is severity of the features. The hair in Fried syndrome tends to be sparser, but the nails in Witkop syndrome are more severely involved. Of course, inheritance pattern also is useful. If successive generations are affected, dominant inheritance and the Witkop syndrome is the likely diagnosis. If parents are normal and one or more of their children are affected, recessive inheritance and the Fried syndrome is the likely diagnosis. Remember, it is hard to determine a pattern of inheritance when only a single child is affected; he or she could have received two copies of a recessive gene from carrier parents or could represent a mutation for a dominant gene. (This complication in the issue of inheritance is mentioned here only as a warning not to be read too much into a family history without guidance from a Geneticist).

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