ECTRODACTYLY-ECTODERMAL DYSPLASIA-CLEFTING (EEC) SYNDROME

The Ectrodactyly-Ectodermal Dysplasia-Clefting syndrome is also known as the EEC syndrome. The word ectrodactyly is derived from the Greek, and means congenital absence of all or part of a finger or toe. While both hands and both feet are usually involved, exceptions have been noted in that one hand may be normal and, rarely, all extremities are normal. Tear ducts are abnormal in most affected individuals; as a result, excessive tearing, inflammation of the eyelids, cornea, and conjunctiva, and sensitivity to light (photophobia) may occur. Skin and hair may be more lightly pigmented than normal and scalp hair, eyebrows, and eyelashes are sparse. Nails in some, but not all, people with the disorder may be abnormal.

The most obvious abnormalities of the mouth include cleft lip associated with cleft palate; cleft of the lip may occur on one or both sides of the lip. In some affected individuals, cleft palate occurs without cleft lip and, rarely, clefting is absent. Congenitally absent teeth and conically shaped teeth are common.

Hearing loss of the conductive type has been noted. In addition, an abnormal voice quality has been observed in some individuals. Additional findings in some affected individuals include abnormalities of the genital urinary tract, choanal atresia, recurrent infections and growth hormone deficiently.

In contrast to some other Ectodermal Dysplasia syndromes in which the inheritance pattern is known, the inheritance pattern in the EEC syndrome is difficult to determine. In most families, only one individual is affected, so one is hard pressed to determine whether parents of the affected child are at risk for having another affected offspring. In other families, transmission of the condition from an affected parent to child has been noted; in these instances, the condition is likely inherited as an autosomal dominant disorder and parents have a 50% chance of giving birth to another affected child with each future pregnancy. Still other reports in the medical literature describe normal parents who have given birth to more than one affected child, suggesting autosomal recessive inheritance; in these instances, parents have a 25% chance of having another affected child. Prenatal diagnosis may be possible by transvaginal ultrasound (looking for structural defects with ultrasound).

There may also be considerable variation in expression between affected members of the same family; that is, some members may have the condition in mild form, while relatives are more severely affected.

Thus, as in any disorder, careful examination of the affected individual, as well as (at least) his or her brothers, sisters and parents is essential before genetic counseling can be accurately provided. There is evidence that the responsible gene, in some families is located on chromosome 7.

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