CONGENITAL ECTODERMAL DYSPLASIA*

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THREE cases of congenital ectodermal dysplasia have been encountered in schools for partially sighted children in the London area. The features of this syndrome are widespread and include the following:

Scarcity of hair growth, which is most conspicuous on the scalp;

Abnormality or absence of the teeth and nails;

Absence of the mucous glands of the nose leading to chronic rhinitis, and dryness of the skin;

Skeletal abnormalities, including depression of the bridge of the nose and underdevelopment of the lower jaw.

The ocular defects associated with the condition are congenital cataracts, which were originally present in two of these cases, and were removed by needlings. In the other case chorioretinal disturbances are present.

Two out of the three cases are females, which is unusual, since it is rare for females to be severely affected by this disorder.

FIG. 1.—Case 1, profile.

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The title given to the disorder is not entirely satisfactory since some tissues of mesodermal origin are involved.

The three cases reported below have the following features in common: fair, dry skin, scanty fair hair, blue eyes, and retarded skeletal growth. There was no consanguinity and no other members of the families were found to be affected.

The disorder has been considered to be due to a genetic anomaly. The mode of inheritance was considered by Cockayne (1933) to be of two types: sex-linked recessive and dominant. Halperin and Curtis (1942) considered the condition to be due to a sex-linked recessive gene with variable expression. Spira (1947) considered the condition to be a manifestation of chronic fluorine poisoning; the addition of fluorides to drinking water as a measure to reduce the incidence of dental caries is of interest in this connection.

Case Reports

(1) A female, aged 7 years, is the youngest of three children, the others being normal in all respects. Her general physique is poor and her height 3 ft. 10 in., a low average for her age. Her intelligence and character are well above average. There is very scanty scalp hair, which, where present, is fair in colour. The eyebrows are absent. The skin is smooth but dry and a chronic rhinitis is present. The nails are normal. The lower jaw is extremely ill-developed (Fig. 1), and the teeth though numerous are dwarfed,

FIG. 2.—Case 2, syndactyly and alopecia.

whilst the fontanelle is still not closed. This latter abnormality makes it necessary to exercise considerable care to avoid injuries to her head, as even ordinary falls are liable to give rise to moderately severe circulatory disturbance.

Both eyes are aphakic, as the result of needlings for congenital cataract. Vision in the right eye is perception of light only and in the left eye 6/36 with + 8 D sph./+ 2.5 D/cyl. axis 90°. Good gaps in the capsule allow adequate fundus examination, which fails to reveal any cause for the poor acuity in the right eye, which admittedly is slightly convergent.

(2) A female, aged 10 years, is an only child. She is stockily built, height 3 ft. 11½ in., well below the average for her age. She walks with a limp, a legacy of Perthe's disease of the right hip, and her hands, but not her feet, show partial syndactyly (Fig. 2, previous page).

The nails are normal. The skin is dry and rough. The scalp hair is fair and scanty with a bald patch over the vertex. Her intelligence appears of low average. Both eyes are aphakic, having been operated on for cataracts, and there is a good view through gaps in the lens capsule of both fundi, which appear normal. Vision in the right eye is amblyopic, only finger counting, whilst in the left eye the visual acuity is 6/60 with + 10 D sph.

(3) A male, aged 9 years, is the third of a family of four children. The parents are not consanguineous and the other members of the family do not show any similar defect. In general appearance the boy shows a marked resemblance to Case 2. His general development is very retarded, height 3 ft. 6 in., compared with the average of 4 ft. 2 in. for a child of that age.

FIG. 3.—Case 3.

His hair and complexion are fair (Fig. 3). The skin is dry; he sweats very little and his mother reported that he has always been very sensitive to cold. Progress in walking and talking was slow compared with that of the other children in the family.

The eyelashes show pili torti, being extremely long and curved.

The vision is 6/60 in each eye with correction:

R	-5.5 D/sph.	L	$-3.5/-4$ D/cyl.
	-2.5 D/cyl.		$\rightarrow 180^\circ$.
	$\rightarrow$ axis 180° .		

The lenses are clear. The fundi show patches of chorioretinal atrophy (Fig. 4), mostly confined to the central areas, the edges being ill-defined, with a few patches at the periphery.

FIG. 4.—Case 3, left fundus. The right fundus presented a similar appearance.

Records show that tests for toxoplasmosis carried out in 1950 were found to be negative. The Wassermann reaction is negative. The lesions are not so clear-cut as in the typical 'colobomatous' type of defect found in toxoplasmosis. The child's mother is also moderately myopic.

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