THE HAND AS A SIGN-POST TO OCULAR DISEASE—II

Hereditary and Developmental Disorders—continued

FIG. 1.—Mongolism.

Mongolism (Down's syndrome)

This is commonly associated with a trisomy of chromosome 21, although more rarely a translocation is present. It is characterized by a typical facies, short stature, mental deficiency, hypogenitalism, and cardiac anomalies. The hands are short, broad, and flabby, with shortening of the thumb. The little fingers show incurving, shortening, and often the absence of one transverse skin crease. The palm, which has an abnormal pattern of tri-radial, also shows a single transverse skin crease.

The typical ocular features include a light-coloured iris with peripheral pallor and Brushfield's spots; the lens is often cataractous, and keratoconus has been reported.

FIG. 2.—Gargoylism.

Gargoylism (Lipochondrodystrophy; Hurler's syndrome)

This is a genetically determined disorder of connective tissue with an excessive production of mucopolysaccharides. The characteristic features consist of the typical facies, mental retardation, skeletal changes, and hepatosplenomegaly. The hand shows limited extensibility of the joints and shortening of the fingers, with incurving of the 4th and 5th digits.

The chief ocular defect, which occurs in 75 per cent. of patients, consists of corneal clouding, which on slit-lamp examination is found to be due to numerous greyish opacities situated in the anterior stromal layers. These opacities are due to the deposition of mucopolysaccharides in the cornea.

Tuberous Sclerosis (Bourneville's disease; Epiloia)

This hereditary defect is characterized by the triad of mental deficiency, epilepsy, and adenoma sebaceum of the face. The affected individuals are predisposed to various tumour formations which include fibromata of the skin, rhabdomyomata of the heart, and gliomata of the central nervous system. The hands may show typical para-ungual fibromata.

The main ocular finding is the presence of large white tumour masses in the retina which may be multiple and mulberry-shaped, and which consist of an overgrowth of glial and embryonic cells with a marked tendency to calcium and hyaline deposition.

FIG. 3.—Tuberous sclerosis.

Neurofibromatosis (von Recklinghausen's disease)

This condition, with a dominant inheritance, is characterized by *café-au-lait* spots on the skin and a wide dissemination of neurofibromata, which occur particularly in the skin, bone, central nervous system, and chromaffin tissue. The hands may demonstrate the *café-au-lait* patches and also the multiple neurofibromata, which appear either as firm subcutaneous nodules or as small violaceous nodules which collapse on pressure.

The ocular manifestations arise from the presence of neurofibromata which may occur in the cornea, iris, choroid, or retina. When they occur extensively in the upper lid (plexiform neuroma) there is an increased incidence of congenital glaucoma. Pulsating exophthalmos may occur and is associated with a deficiency of the posterior orbital wall, while gliomata, particularly of the optic nerve, are not infrequently seen in these patients.

FIG. 4.—Neurofibromatosis.

Illustrations:

THE HOSPITAL FOR SICK CHILDREN, LONDON, W.C.1. (Figs 1 and 2).
GUY'S HOSPITAL, LONDON, S.E.1. (Fig. 4).

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