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

The ophthalmologist derives many clues about associated systemic disease from studying the faces and hands of his patients, and in this and other articles an endeavour will be made to correlate ocular disease with changes in the hands.

Hereditary and Developmental Disorders

Arachnodactyly (Fig. 1)

FIG. 1.—Arachnodactyly.

Marfan's syndrome is an hereditary hypoplastic disorder of mesodermal tissue characterized by ectopia lentis, skeletal and cardiac anomalies, and associated with a defect of collagen causing an elevated urinary hydroxyproline content. Arachnodactyly is diagnosed when the metacarpal index (length/width of metacarpals II-V) is greater than 8.4. It is also found in association with homocystinuria, and in some patients with homozygous (SS) sickle-cell disease.

The main ocular defect, which occurs in almost every case, is ectopia lentis, though microphakia and spherophakia, miosis due to hypoplasia of the iris, and mesodermal dysgenesis, are also found. Abnormalities of the cornea, sclera (blue sclerotics), and retina have also been reported.

FIG. 2.—Brachydactyly.

Brachydactyly (Fig. 2)

Marchesani's syndrome is a hyperplastic mesodermal dystrophy and is thus in direct contrast to Marfan's syndrome. Affected individuals are usually under 5 ft. in height, with a well-developed musculature, brachycephaly, short limbs, broad and short hands, and diminished mobility of the joints.

The ocular defects consist of microphakia and spherophakia with ectopia lentis, and glaucoma may develop with the lens *in situ* or after its dislocation.

Polydactyly (Fig. 3)

The Laurence-Moon-Biedl syndrome is characterized by an atypical pigmentary degeneration of the retina in association with hypogenitalism, obesity, and mental retardation. A similar systemic syndrome with polydactyly, and coloboma of the iris as the ocular defect, has been described (Biemond's syndrome), and the association of bilateral colobomata of the macula with apical dystrophy of the extremities (Sorsby's syndrome) is also rarely seen. More recently, trisomy of chromosomes 13-15 has been found in cases with polydactyly, colobomata, and multiple congenital defects.

FIG. 3.—Polydactyly.

Syndactyly (Fig. 4)

Apert's syndrome (acrocephalo-syndactyly) is characterized by syndactyly with oxycephaly and is due to a mesodermal defect occurring at about the 8th week of embryonic life. The main ocular defects are proptosis, often with exotropia, ophthalmoplegia, and visual failure.

Turner's syndrome of ovarian dysgenesis is characterized by webbing of the neck, renal and skeletal disorders, and coarctation of the aorta. The hand may show syndactyly, with abnormal abduction of the little finger, and the ocular defects include epicanthus, hypertelorism, and oculomotor nerve palsies.

Defects of the limbs are seen in association with congenital heart lesions, and there also appears to be a particular prevalence for the triad of ocular, renal, and skeletal anomalies.

FIG. 4.—Syndactyly.

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