Ophthalmological synopses

Intermediate sex-linked inheritance

The heterozygous state in recessively transmitted conditions can now sometimes be detected as a result of detailed clinical and biochemical investigations. When this occurs in X-linked recessive conditions it is known as *intermediate sex-linked inheritance*, the homozygous (affected) males showing the classical features of the disease and the heterozygous (carrier) females demonstrating minor disorders. Three examples of disorders of the ocular fundus illustrate this point. They are important both from the theoretical point of view and in genetic counselling.

Choroideremia

This is a progressive condition, first appearing in males at or soon after birth and giving rise to pigmentary disturbances in the fundus. As the pigment progressively atrophies the choroidal vessels become visible and then gradually disappear, so that by middle age the sclera is exposed (Fig. 1).

FIG. 1 *Choroideremia*

FIG. 2 *Choroideremia carrier*

In female carriers an irregular and variable peripheral pigmentation of the fundus is present with a cobble-stone type of retinal disturbance (Fig. 2). Occasionally these changes, which are stationary, are similar to those in affected males, but vision is seldom affected.

Sex-linked pigmentary retinal dystrophy

This progressive condition develops in the male in childhood and is characterized by widespread "bone-corporuscle" pigmentation of the fundus, attenuated retinal vessels, and the yellow optic atrophy peculiar to this condition (Fig. 3, opposite).

The classical change in carrier females is a glistening tapetal reflex around the macula. Occasionally the peripheral fundus shows pigmentary mottling and tiny glistening yellowish-white spots (Fig. 4). Visual functions are unimpaired.

FIG. 3 *Sex-linked pigmentary retinal dystrophy*

FIG. 4 *Sex-linked pigmentary retinal dystrophy carrier*

FIG. 5 *Ocular albinism*

FIG. 6 *Ocular albinism carrier*

Ocular albinism

The fundus in affected males is depigmented because of a defect in the formation of melanin, and the choroidal and retinal vessels are clearly visible against a pale background. The foveal reflex is absent (Fig. 5).

In carrier females clumps of pigment, separated by depigmented areas, occur in the periphery of the fundus (Fig. 6).

ILLUSTRATIONS:
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