CASE NOTES

DEGENERATIO PUNCTATA ALBESCENS*

BY

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DEGENERATIO punctata albescens presents one of the most rare and fascinating pictures in ophthalmology, the chief characteristic being the powdering of the entire fundus by discrete white spots. Between the early description by Mooren (1882) and 1939, some fifty cases have been reported in the literature. Other suggested names are "retinitis punctata albescens", "retinosis punctata albescens", and "fundus albi-punctatus cum hemeralopia congenita" (Duke-Elder, 1940).

The present case is the first to be recorded in Egypt, and it throws light on the debatable relationship of this condition to retinitis pigmentosa (Elwyn, 1946). The clinical picture is typical with early onset, stationary course, and positive family history.

Case Report

A male Arab aged 21 years, of dark complexion, complained of non-progressive night blindness since early childhood.

Visual Acuity.

Right eye 6/12, and 6/6 with -0.25 D sph., -0.25 D cyl., axis 90° .

Left eye 6/9, and 6/6 with -0.25 D sph.

Pupillary Reactions.—Sluggish.

Fundus.—Throughout the whole fundus, both centrally and peripherally, were scattered hundreds of sharply-demarcated, dead-white, pigment-free dots. Their shape was mostly rounded, sometimes oval, and occasionally in dumb-bell or hour-glass formation. Their size was about that of retinal vessels of the first or second degree but sometimes larger up to double or triple that size. The distribution was fairly even all over the fundus, starting from near the disc with only minimal affection of the macular area. The spots increased in size and frequency from the centre towards the periphery, so as to cover almost the whole fundus at the periphery.

The retina in between the dots was always normal. The dots lay deeply in a plane behind the retinal vessels. No pigment cells or dots of any shape could be detected anywhere else in the fundus. The retinal veins and arteries were normal, and no sheathing by pigment could be elicited along the course of veins. The macula was normal apart from minimal affection by minute dots at the periphery of the macular area. The foveal reflex was normal but not brightly shining.

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Other Features.—The optic disc, cornea, iris, lens, and vitreous, ocular tension, and colour vision were all normal.

No other congenital malformations were found in the eye or elsewhere in the body.

It is worth mentioning that these dots could be seen by indirect ophthalmoscopy because of their large size, especially in the periphery, and that they differed in this respect from the other cases described.

Visual Fields.—These showed a slight generalized concentric contraction more marked nasally, and this was more marked in a dim light.

Laboratory Investigations.—The Wassermann reaction and Kahn test were both negative. The tuberculin test was negative to 1/100 and 1/10000.

Urine and stools showed nothing abnormal.

The blood cholesterol was 220 mg., and the icterus index 5 units.

Septic foci had been associated with bilateral chronic otitis media since early childhood.

Family History.—This is of outstanding significance. The disease was evidently inherited from the mother, who also suffered from night blindness, and the patient's elder brother, aged 22 years, was also affected. There was no consanguinity in this marriage. The patient's father married three wives after the mother's death. The 2nd wife (who was a blood relation) had a boy aged 15 years, the 3rd wife a girl aged 14 years, and the 4th wife had a boy aged 8 years and six girls aged 9, 7, 6, 5, 4, and 3 years. All these children were unaffected. Further detailed studies of the genetic history of this family are being carried out.

Discussion

The clinical picture with positive family history, very early onset, and non-progressive course establishes the diagnosis of retinitis punctata albescens. Congenital syphilis showing "pepper and salt" with pigment spots of various shapes and large yellowish areas at the periphery (Wolff, 1959), must be considered in the differential diagnosis. In the case described above no pigmentation was seen, and the Wassermann reaction was negative (Tadros, 1955).

Central guttate choroidal atrophy (Tay's choroiditis) with minute yellowish-white spots in the macular area (Troncoso, 1937) should also be considered, but this case showed dead white spots both at the periphery and in the centre of the fundus. Diabetic and albuminuric retinopathies can also be excluded by such typical appearance.

Conclusion

The family history clearly proves the familial incidence and probable recessive mode of inheritance. This does not always show in successive generations; it was not definitely positive in the mother, but occurred in the two brothers (Bishay, 1953). This agrees with Nettleship's observations (Elwyn, 1946): in the first family four children from the first wife were all healthy, and in the second family two sisters were affected out of seven members.

The early onset and non-progressive course, the normal disc and vessels, the complete absence of any pigment cells or sheathing of veins, the normal central vision and the minimal visual field contraction discount any connexion with retinitis pigmentosa (Sobhy, 1949). The patient's dark complexion supports the observations of Cohen (1916) of the occurrence of retinitis punctata in Negroes, and opposes the hypothesis that it is related to retinitis pigmentosa.

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