SUMMARY

Retinitis pigmentosa (RP) is the name commonly given to a group of heredofamilial diseases characterized by progressive visual field loss, night blindness, and abnormal or nonrecordable electroretinogram (ERG). This broad definition encompasses a large number of primary (ocular only) and secondary (other organ or systemic involvement) diseases.

In this thesis, the salient historic, genetic and clinicopathologic aspects of retinitis pigmentosa were outlined.

In addition to the review of literature, the other goal of this thesis was to clinically study the retinitis pigmentosa disease with its different types.

Up to our knowledge, RP patients constituting this series were the first patients with this disease to be subjected to examination by fluorescein angiography in Egypt.

The population of this study included 30 unrelated patients having retinitis pigmentosa examined at Alexandria Eye Institute (26 patients) and Zagazig University Hospital (4 patients).

These patients including parents, siblings and offsprings were examined to determine the hereditary pattern of the disease.

RP patients included in this study were subjected to detailed history taking, complete ophthalmological examination, fundus photography and fluorescein angiography.

For evaluating the value of fluorescein angiography in identifying the disease in patients with no eye ground changes, though at high risk for the development of RP, an additional case was seen in Alexandria Eye Institute.

Among the 30 RP patients (probands), the family history of 7 was negative and indicated no specific hereditary mode, those were termed simplex (s). They constituted 23.4% of the study population, the second largest group. Those with a definable hereditary transmission included 19 patients (63.3%) autosomal recessive (AR),2 patients (6.7%) autosomal dominant (AD), and one patient (3.3%) sex-linked (XL). Only one case fitted the undetermined genetic category (ug).

The male to Female ratio was 2: 1 with a male incidence of 66.7%. Male incidence was higher than female among all genetic groups.

The age of RP patients ranged from 18-53 years with a mean age of 34.56±9.11 years. Out of the 30 patients in this series, 56.7% were more than 32 years old.

Parental consanguinity was observed in 66.7% of RP patients and in 100% of patients in AR group.

Night blindness was the first and main symptom to be complained of by all RP patients in this thesis.

As regards the age of onset of symptoms, 96.7% of patients became symptomatic by the age of 20 years.

Affected visual acuity (/18) amounted to 71.7% in this thesis. There was a statistically significant correlation between the preservation of visual acuity of 6/18 or better and age group. seventy percent of RP patients having more than 32 years of age had corrected visual acuity of less than 6/18.

Myopia was the most prevalent ametropic condition (60%). Of the myopic eyes, 66.7% were less than -6 dioptre.

The spherical refractive error ranged from +4.25 to -12 dioptre.

disease. The presence of retinal pigmentation was a constant finding in all patients. The density of retinal pigmentation was categorized into mild (3.3%), moderate (80%) and severe (26.7%). The type of retinal pigments seen were mainly in the form of bone corpuscles in addition to the less frequent types of rounded, irregular and patchy pigmentation. Whenever there was a central retinal pigmentation, there was always associated peripheral retinal pigmentation. The degree of retinal pigmentation was a measure of the severity of the disease process.

Retinal vessel attenuation was present in all investigated eyes. The degree of retinal vessel attenuation was categorized into mild (6.7%), moderate (50%) and severe (43.3%). There was a statistically significant relationship between the degree of retinal vessel attenuation and increase in age of RP patient, and density of retinal pigmentation, colour of optic disc and degree of visual field affection.

Also, sheathing of the retinal vessels was observed in a number of patients with characteristically advanced disease.

Macular changes were evaluated by both ophthalmoscopy and fluorescein angiography. There was one or more macular changes in 96.7% of studied eyes. The most prevalent macular change was absence of foveal reflex (83.3%). Enlarged macular hypofluorescence (56.7%) and macular atrophy (53.3%) were the next most frequently seen

abnormalities. In all cases with enlarged macular hypofluorescence, the perifoveal capillary bed was not clear.

Other important macular changes were macular pigmentation (13.3%), Bull's eye appearance (6.6%), striate change (preretinal membrane) (23.3%), and macular oedema in 6.6% of eyes.

Peripapillary changes, more evident on fluorescein angiograms than on fundus photographs, represented a constant finding in all RP patients; this observation was not mentioned before in literature.

The most common extraocular association was deafness (13.3%), whereas Usher's syndrome was present in 6.6%. Also 2 patients had a history of diabetes mellitus and non of them showed proliferative diabetic retinopathy.