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Frequency and Clinical Presentation of Retinal Dystrophies - A Hospital Based Study Pak J Ophthalmol Oct 2002;18(4):106-10.

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To study the frequency and clinical features of various types of retinal dystrophies in our population, a prospective hospital based study was conducted at the department of ophthalmology Dow Medical College and Sindh Government Lyarl General Hospital Karachi, Pakistan. Seventy-five patients were diagnosed to have retinal dystrophies out of 60,000 patients who attended the ophthalmic outpatient department from June 1997 to May 2000. One patient out of 800 was found to have some kind of retinal dystrophy. Sixty (80%) patients were under the age of 40 years. There was a male preponderance with 50 (66.7%) males and 25 (33.3%) females. Family history was negative in 48 (64%) patients.

Consanguineous marriages were reported in 43 (57.3%) patients. Different kinds of refractive errors were found In 48 (64%) patients. Commonest retinal dystrophy was Retinitis Pigmentosa (RP). It was diagnosed in 48 (64%) patients. The mode of inheritance was noted to be autosomal recessive In 25 (52%) patients with RP. Two patients had features of Usher's syndrome. Atypical RP was noted in 4 patients. Stargardt's disease was the second common dystrophy identified In 11 (14.7%) cases. Five (6.7%) patients had Cone dystrophies. Final diagnosis could not be ascertained in five (6.7%) patients. This study gives a general idea of pattern of retinal dystrophies in Pakistan. The pattern of retinal dystrophies was found to be similar to the published literature of the western world.