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RETINAL DYSTROPHIES: LANDSCAPE OF GENETIC MUTATIONS IN THE INDIAN SUBCONTINENT

Oral

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Purpose:

To describe the genetic mutations affecting individuals with Inherited Retinal Dystrophies (IRDs), at a tertiary care facility in the Indian Subcontinent

Methods:

Patients attending the Ophthalmology Department of tertiary care hospital and diagnosed with IRD underwent detailed eye exam, retinal imaging included fundus photography and optical coherence tomography. The blood samples were sent for genetic sequencing, specifically clinical exome sequencing to determine the causative mutation. A family history and pedigree analysis was done

Results:

Overall 28 patients underwent genetic sequencing. The common mutations found were in the genes USH2A, CERKL and PROM1. Other mutations seen were IMPG2, PDE6A, CHM, CYP4V2, PCARE, SEMA4A, VPS13B, CABP4, LCA5, and EYS genes. Age of presentation ranged from 5 to 45 years, with the average age 28 years.

Conclusions:

Subset of mutations affecting patients from the Indian Subcontinent tend to be very different from those reported in western literature and elsewhere. A high degree of consanguinity in family members increases the chances of development of IRDs with particular subset of mutations been seen more commonly in certain communities

Mutation Type	GENE	MUTATION one	EXON for MUT one
Del	**CERKL**	**c.1045_1046del; (p.Met349ValfsTer20)**	**exon 8**
Transition	PROM1	c.1946C>T; (p.Ser649Leu)	exon 18
Transversion	MERTK	c.2119A>T; (p.Ile707Phe)	exon 16
Transition	**USH2A**	c.4222C>T, (p.Gln1408Ter)	Exon 19
Del	**CERKL**	**c.967_968delAT p.Met323fs**	**exon 8**
Transversion	**CHM**	**c.820 G>C**	**splice acceptor site**
	IFT 140	c.2686G>A (p.Asp896Asn),	Exon 21
Del	**CERKL**	**c.1045_1046del; (p.Met349ValfsTer20),**	**exon 8**
Transversion	**IMPG2**	c.3423T>G (p. Ser1141Arg)	exon 17
Transition	**PROM1**	c.730C>T, (p.Arg244Ter)	exon 8
	PDE6A	**c.769C>T (p.Arg257Ter)**	**Exon 4**
Del	**CDHR1**	1 bp del c.1459del	exon 10
transition	USH2A	c.13576C>T, (p.Arg4526Ter)	Exon 63
Transition	PROM1	c.2373+2T>C	splice site variant intr
Transition	**CHM**	**c.808 C>T**	**exon 6**
Transversion	**CYP4V2**	c.197T>G (p.Met66Arg)	Exon 1
	PCARE	c.1273C>T (p.Arg425Ter)	exon 1
	SEMA4A	c.1466T>C (p.Val489Ala)	exon 12
	USH2A	c.5012G>A (p.Gly1671Asp)	exon 25
	USH2A	c.5012G>A (p.Gly1671Asp)	exon 25
Del	VPS13B	c.(762+1_763-1)_(2824+1_2825-1)del - (exo	exon 7 - 19
Transition	**CYP4V2**	c.1178C>T, p.Pro393Leu,	Exon 9
Del	CABP4	c.800-2_800-1del (3' splice site)	Intron 5
Transition	PROM1	c.1946C>T; (p.Ser649Leu)	exon 18
Transition & Trasve	**IMPG2**	**c.970G>C, (p.Asp324His)**	**exon 10**
Del	LCA5	c.1151del, (p.Pro384GlnfsTer18)	Exon 8
Transition	EYS	c.9083A>G, (p.Asp3028Gly)	Exon 44
Del	**CERKL**	**c.1045_1046del; (p.Met349ValfsTer20)**	**exon 8**