

(A) Variant zygosity for each diagnostic group associated with typical foveal hypoplasia

Diagnosis	Variant zygosity			
	Compound heterozygous	Homozygous	Hemizygous	Heterozygous
<i>Albinism</i>	149	77	48	55
<i>PAX6</i>	0	0	0	106
<i>SLC38A8</i>	17	16	0	0
<i>AHR</i>	0	2	0	0
<i>FRMD7</i>	0	0	11	6

(B) Type of mutation for each diagnostic group associated with typical foveal hypoplasia

Diagnosis	Variant				
	missense	nonsense	in_del	splice	CTE
<i>Albinism</i>	234	18	61	16	0
<i>PAX6</i>	35	16	38	12	5
<i>SLC38A8</i>	20	7	4	2	0
<i>AHR</i>	0	2	0	0	0
<i>FRMD7</i>	8	1	2	6	0

Diagnosis	Variant (Allele 2 in compound heterozygous cases)			
	missense	nonsense	in_del	splice
<i>Albinism</i>	117	9	17	6
<i>SLC38A8</i>	6	2	5	4