

Person ID	Ocular Phenotype	Systemic Findings	Family History	SOX2 gene	CHX10 gene
012A	Anophthalmia OS, Coloboma OD		None	c.*469C>A ¹	
013A	Bilateral microphthalmia	Cleft lip and palate. Developmental delay.	Adopted	c.*469C>A ¹	
018A	Microphthalmia OS		None		c.471 C>T ² , p.S157S
023A	Microphthalmia with coloboma		None		
024A	Microphthalmia with coloboma OD		None		
025A	Unilateral microphthalmia		None		c.471 C>T ² , p.S157S
027A	Bilateral microphthalmia	46, XX, der (4) t (2,4) (q31.1;q33) Choanal atresia, congenital heart disease, lung hypoplasia, and developmental delay.	None		c.471 C>T ² , p.S157S
028A	Unilateral microphthalmia		None		
029A	Bilateral microphthalmia		None		c.471 C>T ² , p.S157S
030A	Bilateral microphthalmia		None		c.471 C>T ² , p.S157S
031A	Bilateral microphthalmia	Cleft lip and palate.	None		c.471 C>T ² , p.S157S
033A	Microphthalmia OS		None		
034A	Microphthalmia OD		None		
035A	Microphthalmia		None		c.471 C>T ² , p.S157S
037A	Bilateral anophthalmia	Delayed motor and mental development.	None		
038A	Bilateral anophthalmia	Non-syndromic	None		
040A	Bilateral anophthalmia	Dilated ventricles, s/p V-P shunt placement,	None	c.310G>T, (p.E104X)	

		microcephaly. Atrophic optic nerves, chiasm, posterior corpus callosum, and splenium. Abnormal EEG with no overt seizures. Delayed motor and mental development. Short stature. Slightly cupped ears.			
041A	Microphthalmia OS		None		
042A	Bilateral anophthalmia	Duplicated kidney collecting system.	None		c.471 C>T ² , p.S157S
043A	Bilateral microphthalmia	Cleft lip and palate.	None		c.471 C>T ² , p.S157S
044A	Bilateral anophthalmia	Bicuspid aortic valve, atrial septal defect, short stature, growth hormone deficiency, micropenis, bilateral hydrocele, and bilateral cryptorchidism.	None		
045A	Bilateral anophthalmia	Cognitive delay	None		
046A	Anophthalmia OD Microphthalmia and coloboma OD	Motor and developmental delay, “sealed” vagina.	None		
047A	Bilateral microphthalmia	Scimitar syndrome (anomalous pulmonary venous system).	None		
048A	Bilateral anophthalmia	Non-syndromic	Positive-affected maternal cousins, uncles and aunts		
049A	Bilateral anophthalmia	Ventricular-septal defect. Fused teeth.	None	c.*557G>A	c.579 G>A ³ , p.Q193Q

050A	Bilateral anophthalmia		None		
051A	Bilateral microphthalmia	Hypotonia, tremors, lax ligaments.	Mother with polar cataracts		
052A	Bilateral microphthalmia		Sister to 52D		
052D	Colobomas OU		Sister to 52A		
054A	Microphthalmia OS Anophthalmia OD	Partial complex seizures, right middle fossa arachnoid cyst and partial absence of posterior aspect of corpus callosum including the splenium. Mild hydrocephalus. Growth and thyroid hormone deficiency. Developmental delays. Wide-based ataxic gait. Facial asymmetry with beaked nose, small widely spaced teeth, diastasis recti. Hypoplastic labia minora.	Sister to 54H	c.549delC, (p.P184Rfs)	
054H	Bilateral anophthalmia by ultrasound <i>in utero</i>	Partial agenesis of the corpus callosum. Growth and thyroid hormone deficiency. Developmental delays.	Sister to 54A	c.549delC	c.471 C>T ² , p.S157S
055A	Microphthalmia OD Anophthalmia OS		None		c.871 G>A ³ . p.D291N
056A	Bilateral anophthalmia	Developmental delay.	None		