INVITAE OPHTHALMOLOGY GENE PANEL TESTS

Our testing menu is curated by medical genetics experts and covers some common causes of non-syndromic and syndromic ophthalmic conditions, including retinal, lens, and anterior segment dysgenesis disorders.

CLINICAL AREA: OPHTHALMOLOGY

Test name	# gene(s)	Gene list
Eye Disorders		
Invitae Aniridia Test	1	PAX6
Invitae Axenfeld-Rieger Panel	2	FOXC1, PITX2
Add-on aniridia gene	1	PAX6
Invitae Bardet-Biedl Syndrome Panel	16	ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP290, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP
Invitae CHARGE Syndrome Test	1	CHD7
Invitae Choroideremia Test	1	CHM
Invitae Congenital Cataracts Panel	34	AGK, BCOR, BFSP1, BFSP2, CRYAA, CRYAB, CRYBA1, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGC, CRYGD, CRYGS, CTDP1, EPHA2, FAM126A, FOXC1, FYCO1, GALK1, GCNT2, GJA3, GJA8, HSF4, MAF, MIP, NHS, OCRL, PAX6, PITX2, PITX3, SIL1, TDRD7, VSX2
Add-on preliminary-evidence genes	4	CHMP4B, CRYGB, LIM2, VIM
Invitae Duane-Radial Ray Syndrome Test	1	SALL4
Invitae Early-Onset Glaucoma Panel	3	CYP1B1, FOXC1, PITX2
Invitae Leber Congenital Amaurosis Panel	19	AIPL1, CEP290, CRB1, CRX, GDF6, GUCY2D, IQCB1, KCNJ13, LCA5, LRAT, NMNAT1, OTX2, PRPH2, RD3, RDH12, RPE65, RPGRIP1, SPATA7, TULP1
Add-on preliminary-evidence genes	2	BBS4, IMPDH1
Invitae Microphthalmia/Anophthalmia Disorders Panel	17	ALDH1A3, BCOR, BMP4, FOXE3, GDF6, MAB21L2, MFRP, OTX2, PAX2, PRSS56, PXDN, RARB, RAX, SHH, SOX2, STRA6, VSX2
Add-on preliminary-evidence genes	3	GDF3, HESX1, VAX1
Invitae Oculo-Facio-Cardio-Dental Syndrome Test	1	BCOR
Invitae Retinoblastoma Test	1	RB1
Invitae Senior-Loken Syndrome Panel	8	CEP290, INVS, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, WDR19

