

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: PEDIATRIC AND RARE DISEASE

INHERITED EYE DISORDERS

Invitae Aniridia Test (1 gene)	Primary panel (1 gene) PAX6
Invitae Axenfeld-Rieger Panel (up to 3 genes)	Primary panel (2 genes) FOXC1 PITX2 Add-on aniridia gene (1 gene) PAX6
Invitae Bardet-Biedl Syndrome Panel (16 genes)	Primary panel (16 genes) ARL6 BBS1 BBS10 BBS12 BBS2 BBS4 BBS5 BBS7 BBS9 CEP290 MKKS MKS1 SDCCAG8 TRIM32 TTC8 WDPCP
Invitae CHARGE Syndrome Test (1 gene)	Primary panel (1 gene) CHD7
Invitae Choroideremia Test (1 gene)	Primary panel (1 gene) CHM
Invitae Congenital Cataracts Panel (up to 38 genes)	Primary panel (33 genes) 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 VSX2 Add-on preliminary-evidence genes (5 genes) CHMP4B CRYGB LIM2 TDRD7 VIM
Invitae Duane-Radial Ray Syndrome Test (1 gene)	Primary panel (1 gene) SALL4
Invitae Early-Onset Glaucoma Panel (3 genes)	Primary panel (3 genes) CYP1B1 FOXC1 PITX2
Invitae Leber Congenital Amaurosis Panel (up to 21 genes)	Primary panel (19 genes) 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 genes) BBS4 IMPDH1
Invitae Microphthalmia/Anophthalmia Disorders Panel (up to 20 genes)	Primary panel (17 genes) ALDH1A3 BCOR BMP4 FOXE3 GDF6 MAB21L2 MFRP OTX2 PAX2 PRSS56 PXDN RARB RAX SHH SOX2 STRA6 VSX2 Add-on preliminary-evidence genes (3 genes) GDF3 HESX1 VAX1
Invitae Oculo-Facio-Cardio-Dental Syndrome Test (1 gene)	Primary panel (1 gene) BCOR
Invitae Retinoblastoma Test (1 gene)	Primary panel (1 gene) RB1
Invitae Senior-Loken Syndrome Panel (8 genes)	Primary panel (8 genes) CEP290 INVS IQCB1 NPHP1 NPHP3 NPHP4 SDCCAG8 WDR19

ABOUT INVITAE

Invitae's mission is to bring comprehensive genetic information into mainstream medical practice to improve the quality of healthcare for everyone. Our goal is to aggregate most of the world's genetic tests into a single service with higher quality, faster turnaround time, and lower price than many single-gene tests today.

For more information about Invitae's genetic tests, please visit www.invitae.com.

