

## 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

<b>Invitae Aniridia Test</b> (1 gene)	<b>Primary panel (1 gene)</b> PAX6
<b>Invitae Axenfeld-Rieger Panel</b> (up to 3 genes)	<b>Primary panel (2 genes)</b> FOXC1 PITX2 <b>Add-on aniridia gene (1 gene)</b> PAX6
<b>Invitae Bardet-Biedl Syndrome Panel</b> (16 genes)	<b>Primary panel (16 genes)</b> ARL6 BBS1 BBS10 BBS12 BBS2 BBS4 BBS5 BBS7 BBS9 CEP290 MKKS MKS1 SDCCAG8 TRIM32 TTC8 WDPCP
<b>Invitae CHARGE Syndrome Test</b> (1 gene)	<b>Primary panel (1 gene)</b> CHD7
<b>Invitae Choroideremia Test</b> (1 gene)	<b>Primary panel (1 gene)</b> CHM
<b>Invitae Congenital Cataracts Panel</b> (up to 38 genes)	<b>Primary panel (32 genes)</b> AGK BCOR BFSP1 BFSP2 CRYAA CRYAB CRYBA1 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 <b>Add-on preliminary-evidence genes (6 genes)</b> CHMP4B CRYBA4 CRYGB LIM2 TDRD7 VIM
<b>Invitae Duane-Radial Ray Syndrome Test</b> (1 gene)	<b>Primary panel (1 gene)</b> SALL4
<b>Invitae Early-Onset Glaucoma Panel</b> (3 genes)	<b>Primary panel (3 genes)</b> CYP1B1 FOXC1 PITX2
<b>Invitae Leber Congenital Amaurosis Panel</b> (up to 21 genes)	<b>Primary panel (19 genes)</b> AIPL1 CEP290 CRB1 CRX GDF6 GUCY2D IQCB1 KCNJ13 LCA5 LRAT NMNAT1 OTX2 PRPH2 RD3 RDH12 RPE65 RPGRIP1 SPATA7 TULP1 <b>Add-on preliminary-evidence genes (2 genes)</b> BBS4 IMPDH1
<b>Invitae Microphthalmia/Anophthalmia Disorders Panel</b> (up to 20 genes)	<b>Primary panel (15 genes)</b> ALDH1A3 BCOR BMP4 FOXE3 GDF6 MFRP OTX2 PAX2 PRSS56 PXDN RAX SHH SOX2 STRA6 VSX2 <b>Add-on preliminary-evidence genes (5 genes)</b> GDF3 HESX1 MAB21L2 RARB VAX1
<b>Invitae Oculo-Facio-Cardio-Dental Syndrome Test</b> (1 gene)	<b>Primary panel (1 gene)</b> BCOR
<b>Invitae Retinoblastoma Test</b> (1 gene)	<b>Primary panel (1 gene)</b> RB1
<b>Invitae Senior-Loken Syndrome Panel</b> (8 genes)	<b>Primary panel (8 genes)</b> 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](http://www.invitae.com).

