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|---|---|
| Full name: | Cataract - UGent |
| Type of panel: | in-house |
| Version number: | v3 |
| Laboratory: | Centrum Medische Genetica - UZ Gent |
| Created: | 17 Jun 2019 - 11:49 |
| Changed: | 16 Dec 2022 - 10:38 |

Related Diseases

- 3-methylglutaconic aciduria type 3
- Autosomal dominant cutis laxa
- Autosomal dominant optic atrophy and cataract
- Autosomal dominant spastic paraplegia type 9A
- Autosomal dominant spastic paraplegia type 9B
- Autosomal dominant vitreoretinochoroidopathy
- Autosomal recessive bestrophinopathy
- Autosomal recessive spastic paraplegia type 9B
- Axenfeld-Rieger syndrome
- BOR syndrome
- Branchiootic syndrome
- Cataract-microcornea syndrome
- Cerebrotendinous xanthomatosis
- Cerulean cataract
- Classic galactosemia
- Cockayne syndrome type 1
- Cockayne syndrome type 2
- Cockayne syndrome type 3
- Coloboma of choroid and retina
- Congenital cataracts-facial dysmorphism-neuropathy syndrome
- Coralliform cataract

- Craniometaphyseal dysplasia
- Dent disease type 2
- Early-onset anterior polar cataract
- Early-onset lamellar cataract
- Early-onset non-syndromic cataract
- Early-onset nuclear cataract
- Early-onset posterior polar cataract
- Early-onset posterior subcapsular cataract
- Early-onset sutural cataract
- Erythrokeratodermia variabilis
- Familial exudative vitreoretinopathy
- Familial isolated dilated cardiomyopathy
- Familial multiple meningioma
- Familial thoracic aortic aneurysm and aortic dissection
- Full schwannomatosis
- Galactokinase deficiency
- Hereditary hyperferritinemia-cataract syndrome
- Hypochondrogenesis
- Hypomyelination-congenital cataract syndrome
- Isolated aniridia
- Isolated congenital sclerocornea
- Isolated optic nerve hypoplasia/aplasia
- Kniest dysplasia
- Knobloch syndrome
- Legg-Calvé-Perthes disease
- Microphthalmia, Lenz type
- Morning glory disc anomaly
- Nance-Horan syndrome
- Oculocerebrorenal syndrome of Lowe
- Oculodentodigital dysplasia
- Otofaciocervical syndrome
- Platyspondylic dysplasia, Torrance type
- Pulverulent cataract
- Rare isolated myopia
- Retinitis pigmentosa
- Retinopathy of prematurity

- Stickler syndrome type 1
- Syndactyly type 3
- Total early-onset cataract
- UV-sensitive syndrome

Related Analytes

| GENE | % OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS | COPY NUMBER VARIATION | COMMENTS |
|---|---|---|---|
| ABHD12 | 99.98 | 1 | |
| ADAMTSL4 | 99.66 | 1 | |
| AGK | 99.99 | 1 | |
| ALDH18A1 | 99.96 | 1 | |
| B3GLCT | 99.90 | 1 | |
| BCOR | 99.97 | 1 | |
| BEST1 | 99.86 | 1 | |
| BFSP1 | 100.00 | 1 | |
| BFSP2 | 99.09 | 1 | |
| CHMP4B | 99.96 | 1 | |
| COL11A1 | 90.72 | 1 | |
| COL18A1 | 99.99 | 1 | |
| COL2A1 | 99.87 | 1 | |
| CRYAA | 19.49 | 1 | |
| CRYAB | 100.00 | 1 | |
| CRYBA1 | 99.99 | 1 | |

| GENE | % OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS | COPY NUMBER VARIATION | COMMENTS |
|---|---|---|---|
| CRYBA2 | 100.00 | 1 | |
| CRYBA4 | 100.00 | 1 | |
| CRYBB1 | 99.46 | 1 | |
| CRYBB2 | 99.94 | 1 | |
| CRYBB3 | 99.99 | 1 | |
| CRYGB | 99.99 | 1 | |
| CRYGC | 100.00 | 1 | |
| CRYGD | 100.00 | 1 | |
| CRYGS | 100.00 | 1 | |
| CTDP1 | 99.97 | 1 | |
| CYP27A1 | 100.00 | 1 | |
| CYP51A1 | 97.82 | 1 | |
| DNMBP | 99.94 | 1 | |
| EPG5 | 99.95 | 1 | |
| EPHA2 | 99.99 | 1 | |
| EYA1 | 99.81 | 1 | |
| FBN1 | 99.85 | 1 | |
| FOXE3 | 99.29 | 1 | |
| FTL | 99.99 | 1 | |
| FYCO1 | 100.00 | 1 | |

| GENE | % OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS | COPY NUMBER VARIATION | COMMENTS |
|---|---|---|---|
| FZD4 | 100.00 | 1 | |
| GALK1 | 100.00 | 1 | |
| GALT | 100.00 | 1 | |
| GCNT2 | 100.00 | 1 | |
| GJA1 | 100.00 | 1 | |
| GJA3 | 100.00 | 1 | |
| GJA8 | 99.99 | 1 | |
| HMX1 | 100.00 | 1 | |
| HSF4 | 99.98 | 1 | |
| HYCC1 | 99.81 | 1 | |
| INPP5K | 99.94 | 1 | |
| INTS1 | 100.00 | 1 | |
| JAM3 | 100.00 | 1 | |
| LCAT | 99.97 | 1 | |
| LEMD2 | 100.00 | 1 | |
| LIM2 | 100.00 | 1 | |
| LSS | 99.98 | 1 | |
| MAF | 99.73 | 1 | |
| MIPEP | 99.98 | 1 | |
| MIR184 | 100.00 | 1 | |

| GENE | % OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS | COPY NUMBER VARIATION | COMMENTS |
|---|---|---|---|
| MYH9 | 99.95 | 1 | |
| NDP | 99.98 | 1 | |
| NF2 | 100.00 | 1 | |
| NHS | 99.96 | 1 | |
| OCRL | 99.89 | 1 | |
| OPA3 | 100.00 | 1 | |
| P3H2 | 99.93 | 1 | |
| PANK4 | 100.00 | 1 | |
| PAX6 | 99.95 | 1 | |
| PITX3 | 100.00 | 1 | |
| PXDN | 100.00 | 1 | |
| RRAGA | 100.00 | 1 | |
| SIL1 | 99.95 | 1 | |
| SIPA1L3 | 99.97 | 1 | |
| SLC16A12 | 100.00 | 1 | |
| SLC33A1 | 99.67 | 1 | |
| TDRD7 | 99.87 | 1 | |
| UNC45B | 100.00 | 1 | |
| VIM | 100.00 | 1 | |
| VSX2 | 99.99 | 1 | |

| GENE | % OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS | COPY NUMBER VARIATION | COMMENTS |
|---|---|---|---|
| WFS1 | 99.99 | 1 | |