DISEASE:
Leber congenital amaurosis

NAME:	Leber congenital amaurosis
DESCRIPTION:	Leber congenital amaurosis (LCA) is a retinal dystrophy defined by blindness and responses to electrophysiological stimulation (Ganzfeld electroretinogram (ERG)) below threshold, associated with severe visual impairment within the first year of life.
ORPHACODE:	65
SYNONYMS:	Amaurosis congenita of Leber

XREF(S):	Orphanet MeSH ICD-10 OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM MedDRA OMIM

ANALYTE(S):	USP45 TUBB4B SPATA7 GDF6 IFT140 NMNAT1 RD3 RDH12 RPE65 RPGRIP1 CEP290 AIPL1 TULP1 CRB1 CRX GUCY2D IMPDH1 IQCB1 LCA5 KCNJ13 LRAT PCYT1A
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

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RELATED CONTENT

Related Genetic Tests

- Ciliopathy (gene panel)
- Leber Congenital Amaurosis - Retinal dystrophy, early onset (gene panel)

Related Laboratories

- Centrum Medische Genetica - UZ Gent

Related Analytes

- aryl hydrocarbon receptor interacting protein like 1
- centrosomal protein 290
- crumbs cell polarity complex component 1
- cone-rod homeobox
- growth differentiation factor 6
- guanylate cyclase 2D, retinal
- intraflagellar transport 140
- inosine monophosphate dehydrogenase 1
- IQ motif containing B1
- potassium inwardly rectifying channel subfamily J member 13
- lebercilin LCA5
- lecithin retinol acyltransferase
- nicotinamide nucleotide adenylyltransferase 1
- phosphate cytidylyltransferase 1A, choline

- RD3 regulator of GUCY2D
- retinol dehydrogenase 12
- retinoid isomerohydrolase RPE65
- RPGR interacting protein 1
- spermatogenesis associated 7
- tubulin beta 4B class IVb
- TUB like protein 1
- ubiquitin specific peptidase 45

Related Gene Panels

- Autosomal dominant Retinitis pigmentosa (11 genes) - UGent
- Ciliopathy (120 genes) - UGent
- Hypogonadotropic Hypogonadism/Kallmann (61 genes) - ULG
- Leber - Retinal, early onset
- Leber Congenital Amaurosis - UGent

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