ANALYTE:
GJA1

NAME:	gap junction protein alpha 1
SYMBOL:	GJA1
VERSION OF ORPHANET:	2023-06-22 14:14:43
SYNONYMS:	CX43 ODD ODOD SDTY3 connexin 43 oculodentodigital dysplasia (syndactyly type III)
XREF(S):	Orphanet Ensembl Genatlas HGNC OMIM Reactome SwissProt
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RELATED CONTENT

Related Genetic Tests

- Arteriovenous malformation (gene panel)
- Capillary malformation - arteriovenous malformation (2 genes)
- Cataract (gene panel)
- Cerebral cavernous malformation (gene panel)
- Congenital malformation (gene panel - 1721 genes)
- Congenital malformation gene panel
- Congenital structural heart defects (gene panel)
- Dermatogenetic panel, severe, rare and hereditary genodermatoses (gene panel - 394 genes)
- Glaucoma (gene panel)
- Heart / Cardio disorders / Cardiopathy (gene panel)
- Hereditary spastic paraplegia (gene panel - 249 genes)
- Ichthyosis (gene panel)
- Intellectual disability & Epilepsy (gene panel)
- Intellectual disability (virtual gene panel)
- Leukodystrophy (gene panel)
- Microphthalmia / Anophthalmia / Coloboma-Anterior Segment Dysgenesis (MAC-ASD) (gene panel)
- Oculo Dento Digital Dysplasia
- Primary Electrical disorders / Brugada syndrome / Long QT syndrome (LQT) / Short QT syndrome (SQT) / Arrhythmogenic right ventricular cardiomyopathy (ARVC) / Catecholaminergic polymorphic ventricular tachycardia (CPVT) (gene panel)
- Primary ciliary dyskinesia (PCD) Heterotaxyies (gene panel)
- Primary lymphedema / fetal hydrops (gene panel)
- Rendu-Osler-Weber disease (4 genes)
- Skeletal dysplasia (gene panel)
- Skeletal dysplasia (gene panel)
- Skeletal dysplasia (gene panel)
- Skin disorders (gene panel)

- Spastic Paraplegia (gene panel)
- Venous malformation (3 genes)
- cleft lip with/whitout cleft palate (virtual gene panel)

Related Diseases

- Autosomal dominant palmoplantar keratoderma and congenital alopecia
- Craniometaphyseal dysplasia
- Erythrokeratodermia variabilis
- Hypoplastic left heart syndrome
- Oculodentodigital dysplasia
- Rare autosomal recessive non-syndromic sensorineural deafness type DFNB
- Syndactyly type 3

Related Gene Panels

- Cataract - UGent
- Cleft lip and palate / dysmorphic facial features / craniofacial anomalies (255 genes)) - UCL
- Congenital heart disease (29 genes) - VUB
- Congenital malformation (1721 genes) - ULB
- Congenital malformation gene panel - VUB
- Congenital structural heart defects - UGent
- Dermatogenetic / severe, rare and hereditary genodermatoses (394 genes) - ULB
- Glaucoma - UGent
- Hereditary spastic paraplegia (188 genes) - ULB
- Heterotaxie PCD - UGent
- Ichthyosis and erythroderma (98 genes) - KUL
- Intellectual disability & Epilepsy - UGent
- Intellectual disability (gene panel)
- Leukodystrophy - UGent
- Lymphedema / fetal hydrops (27 genes) - UCL

- Microphthalmia/Anophthalmia/Coloboma - Anterior Segment Dysgenesis - UGent
- Primary Electrical disorders/Brugada syndrome (genepanel) - UZA
- Skeletal dysplasia (394 genes) - VUB
- Skeletal dysplasia (genepanel) - UZA
- Skeletal dysplasia - UGent
- Skin disorders - UGent
- Spastic Paraplegia (89 genes) - IPG
- Vascular malformations (germline) (38 genes) - UCL
- cardiopathy panel - UGent
- test-test

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