Cleft Lip and Palate

Precision Panel

Overview

Cleft Lip and Palate (CLP) are a group of malformations of unknown etiology with overlapping phenotypes affecting both females and males. They are the most common congenital malformations of the craniofacial region. Cleft lip and palate can be an isolated finding (non-syndromic), or they can present as a syndromic form. In general, sporadic cases with no family history are related to environmental risks, while the presence of one or more affected relatives strongly suggests that genetic factors are the main contributor. The etiology is multifactorial including both genetic predisposition and environmental factors. Individuals with cleft lip and/or palate may experience difficulties with feeding, talking, hearing and social integration. Typically, individuals affected with these disorders have a higher morbidity and mortality throughout life than do unaffected individuals.

The Igenomix Cleft Lip and Palate Precision Panel can be used to make a directed and accurate diagnosis of syndromic versus nonsyndromic causes of cleft lip and/or palate ultimately leading to a better management and prognosis of the disease. It provides a comprehensive analysis of the genes involved in this disease using next-generation sequencing (NGS) to fully understand the spectrum of relevant genes involved.

Indications

The Igenomix Cleft Lip and Palate Precision Panel is indicated for those patients with a clinical suspicion or diagnosis presenting with the following manifestations:

- Split in the lip and/or roof of the mouth
- Feeding difficulty
- Difficulty swallowing
- Nasal speaking voice
- Chronic ear infections

Clinical Utility

The clinical utility of this panel is:

- The genetic and molecular confirmation for an accurate clinical diagnosis of a symptomatic patient.
- Early initiation of multidisciplinary treatment including surgical repair and prevention for future pregnancies if actionable.
- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.

- Improvement of delineation of genotype-phenotype correlation.
- Identification of the genetic basis of these associated disorders for a better insight into the mechanisms of craniofacial development.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ABCA12	Multiple Ichthyosis Types	AR	100	151 of 153
ABCA4	Cone-Rod Dystrophy, Macular Degeneration, Retinitis Pigmentosa, Stargardt Disease	AD,AR	100	1392 of 1430
ACACB	Biotin Deficiency, Fatty Liver Disease, Diabetes Mellitus	-	99	9 of 9
ACBD5	Retinal Dystrophy, Leukodystrophy	AR	100	3 of 3
ACSS2	Cleft Lip, Cystoisosporiasis, Cardiomyopathy, Canavan Disease	-	96.99	1 of 1
ACTB	Baraitser-Winter Syndrome, Dystonia, Becker Nevus Syndrome, Deafness	AD	100	40 of 40
ACTC1	Atrial Septal Defect, Cardiomyopathy	AD	99.93	72 of 74
ACTG2	Visceral Myopathy, Megacystis, Microcolon, Intestinal Hypoperistalsis	AD	99.91	23 of 23
ADA2	Polyarteritis Nodosa, Sneddon Syndrome, Blackfan-Diamond Anemia	AR	100	-
ADH7	Gastritis	-	99.87	-
AHDC1	Xia-Gibbs Syndrome, Intellectual Disability, Obstructive Sleep Apnea	AD	99.87	41 of 43
AHI1	Joubert Syndrome, Retinitis Pigmentosa	AR	96.79	85 of 97
ALG3	Congenital Disorder Of Glycosylation	AR	99.2	25 of 25
ALX1	Frontonasal Dysplasia, Cleft Lip Palate	AR	100	3 of 3
ALX3	Frontonasal Dysplasia, Frontorhiny	AR	89.31	8 of 8
ALX4	Craniosynostosis, Frontonasal Dysplasia, Parietal Foramina, Scaphocephaly, Potocki-Shaffer Syndrome	AD,AR	99.94	25 of 25
AMELX	Amelogenesis Imperfecta	X,XD,G	100	-
AMER1	Osteopathia Striata, Cranial Sclerosis	X,XD,G	99.45	-
AMMECR1	Midface Hypoplasia, Hearing Impairment, Elliptocytosis, Nephrocalcinosis, Alport Syndrome, Intellectual Disability	X,XR,G	99.81	-
ANKRD11	Kbg Syndrome, 16q24.3 Microdeletion Syndrome	AD	99.6	119 of 124
ANOS1	Hypogonadotropic Hypogonadism, Kallmann Syndrome	X,XR,G	96.86	-
ARCN1	Short Stature, Rhizomelic, Microcephaly, Micrognathia, Developmental Delay	AD	99.91	4 of 4
ARHGAP29	Cleft Lip/Palate	-	99.87	19 of 19
ARHGAP31	Adams-Oliver Syndrome	AD	100	6 of 6
ARID1A	Coffin-Siris Syndrome	AD	95.32	40 of 42
ARID1B	Coffin-Siris Syndrome, 6q25 Microdeletion Syndrome	AD	93.87	226 of 238
ARID2	Coffin-Siris Syndrome	AD	99.97	17 of 17
ARID5B	Leukemia, Adenoid Cystic Carcinoma	-	99.92	3 of 3
ARNT2	Webb-Dattani Syndrome, Septo-Optic Dysplasia Spectrum	AR	100	5 of 5
ARVCF	22q11.2 Deletion Syndrome	-	99.95	2 of 2
ARX	Corpus Callosum, Epileptic Encephalopathy, Lissencephaly, Mental Retardation, Partington Syndrome, West Syndrome	X,XR,G	81.92	-
ASXL1	Bohring-Opitz Syndrome, Myelodysplastic Syndrome, Mastocytosis, Bohring-Opitz Syndrome	AD	99.96	41 of 41
ATN1	Congenital Hypotonia, Developmental Delay, Dentatorubral-Pallidoluysian Atrophy, Naito-Oyanagi Disease, Ataxia, Chorea, Seizures, Dementia	AD	99.86	11 of 11

ATP6V1B2	Deafness, Onychodystrophy, Zimmermann-Laband Syndrome, Deafness	AD	100	5 of 5
ATR	Cutaneous Telangiectasia And Cancer Syndrome, Seckel Syndrome	AD,AR	99.98	39 of 40
ATRX	Alpha-Thalassemia Myelodysplasia Syndrome, Carpenter-Waziri Syndrome, Chudley-Lowry-Hoar Syndrome, Holmes-Gang Syndrome, Juberg-Marsidi Syndrome, Smith-Fineman-Myers Syndrome	X,XR,XD,G	98.5	-
B3GALNT2	Limb Girdle Muscular Dystrophy, Dystroglycanopathy, Intellectual Disability, Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	97.14	17 of 17
B3GALT6	Ehlers-Danlos Syndrome, Spondyloepimetaphyseal Dysplasia, Joint Laxity	AR	65.09	24 of 39
B3GAT3	Multiple Joint Dislocations, Craniofacial Dysmorphism	AR	99.86	15 of 15
B3GLCT	Peters-Plus Syndrome	AR	99.96	-
B4GALT7	Ehlers-Danlos Syndrome	AR	99.92	11 of 11
B4GAT1	Limb Girdle Muscular Dystrophy, Walker-Warburg Syndrome	AR	-	-
B9D1	Joubert Syndrome, Meckel Syndrome	AR	90.23	11 of 11
B9D2	Meckel Syndrome	AR	84.81	4 of 5
BCOR	Microphthalmia, Promyelocytic Leukemia, Oculofaciocardiodental Syndrome	X,XD,G	99.87	-
BCR	Leukemia, 22q11.2 Microdeletion Syndrome	MU,P	97.78	-
BGN	Meester-Loeys Syndrome, Spondyloepimetaphyseal Dysplasia	X,XR,G	99.87	-
BIN1	Myopathy	AR	100	20 of 20
BMP2	Brachydactyly, Hemochromatosis, Short Stature, Skeletal Anomalies, Cardiac Anomalies, 20p12.3 Microdeletion Syndrome	AD,AR	99.48	12 of 12
BMP4	Microphthalmia, Cleft Lip/Palate	AD,MU,P	100	38 of 42
BMPER	Diaphanospondylodysostosis	AR	99.98	22 of 22
BPNT2	Chondrodysplasia, Joint Dislocations	AR	97.04	4 of 4
BRAF	Cardiofaciocutaneous Syndrome, Leopard Syndrome, Lung Cancer, Noonan Syndrome, Craniopharyngioma	AD	100	80 of 80
BRCA1	Breast And Ovarian Cancer, Fanconi Anemia, Pancreatic Carcinoma, Primary Peritoneal Carcinoma	AD,AR,MU	98.97	2783 of 2894
BRCA2	Breast And Ovarian Cancer, Fanconi Anemia, Glioma, Medulloblastoma, Pancreatic Cancer, Prostate Cancer, Wilms Tumor, Nephroblastoma	AD,AR,MU	98.51	3343 of 3451
BRIP1	Fanconi Anemia, Breast And Ovarian Cancer	AD,AR	94.97	235 of 237
BUB1	Colorectal Cancer, Mosaic Variegated Aneuploidy Syndrome	AD	99.76	18 of 19
BUB1B	Colorectal Cancer, Mosaic Variegated Aneuploidy Syndrome	AD,AR	99.84	30 of 31
BUB3	Mosaic Variegated Aneuploidy Syndrome	-	99.98	6 of 6
C2CD3	Orofaciodigital Syndrome	AR	97.25	18 of 18
CADPS	Jejunal Somatostatinoma, Acrofacial Dysostosis	-	97.52	5 of 5
CARS1	Microcephaly, Developmental Delay, Brittle Hair Syndrome	AR	100	7 of 7
CASK	Anemia, Fg Syndrome, Mental Retardation, Microcephaly, Pontine, Cerebellar Hypoplasia, Epileptic Encephalopathy	X,XR,XD,G	99.98	-
CBFB	Leukemia	AD	100	-
CC2D2A	Coach Syndrome, Joubert Syndrome, Meckel Syndrome	AR	99.43	98 of 100
CCDC141	Hypogonadotropic Hypogonadism, Kallmann Syndrome	AR	99.7	1 of 1
CCDC22	Ritscher-Schinzel Syndrome	X,XR,G	99.94	-
CD96	C Syndrome	AD	100	4 of 4
CDC45	Meier-Gorlin Syndrome, Ear-Patella-Short Stature Syndrome	AR	99.99	19 of 19
CDC6	Meier-Gorlin Syndrome, Ear-Patella-Short Stature Syndrome	AR	100	2 of 2
CDH1	Blepharocheilodontic Syndrome, Breast Cancer, Endometrial Carcinoma, Gastric Cancer, Prostate Cancer, Suppressor Of	AD	100	361 of 363

	Tumorigenicity, Blepharo-Cheilo-Odontic Syndrome, Cleft Lip/Palate			
CDH11	Elsahy-Waters Syndrome, Branchioskeletogenital Syndrome	AR	99.95	10 of 10
CDH19	Charge Syndrome, Mocrophthalmia, Coloboma	-	98.64	2 of 2
CDKL5	Epileptic Encephalopathy, Rett Syndrome, West Syndrome	X,XD,G	99.92	-
CDKN1C	Beckwith-Wiedemann Syndrome, Intrauterine Growth Retardation, Metaphyseal Dysplasia, Hypoplasia, Genital Anomalies, Image Syndrome, Diabetes	AD	73.58	55 of 76
CDON	Holoprosencephaly, Pituitary Stalk Interruption Syndrome	AD	100	15 of 15
CDT1	Meier-Gorlin Syndrome, Ear-Patella-Short Stature Syndrome	AR	97.43	12 of 12
CENPF	Stromme Syndrome	AR	98.83	10 of 12
CEP120	Joubert Syndrome, Short-Rib Thoracic Dysplasia, Jeune Syndrome	AR	99.8	9 of 9
CEP290	Bardet-Biedl Syndrome, Joubert Syndrome, Leber Amaurosis, Meckel Syndrome, Senior-Loken Syndrome	AR	96.47	293 of 327
CEP41	Joubert Syndrome	AR	100	17 of 17
CEP55	Multinucleated Neurons, Renal Dysplasia, Cerebellar Hypoplasia, Hydranencephaly, Meckel Syndrome	AR	99.22	3 of 3
CEP57	Mosaic Variegated Aneuploidy Syndrome	AR	99.64	6 of 6
CFDP1	Inflammatory Bowel Disease	-	98.99	1 of 1
CHD7	Charge Syndrome, Hypogonadotropic Hypogonadism, Kallmann Syndrome, Omenn Syndrome	AD	96.25	823 of 896
CHN1	Duane Retraction Syndrome	AD	92.41	11 of 11
CHN2	Schizophrenia, Neuropathy Of Dejerine-Sottas	-	99.94	-
CHRNA1	Multiple Pterygium Syndrome, Myasthenic Syndrome	AD,AR	100	35 of 35
CHRNA7	15q13.3 Microdeletion Syndrome	AD	82.09	2 of 2
CHRND	Multiple Pterygium Syndrome, Myasthenic Syndrome	AD,AR	100	31 of 31
CHRNG	Multiple Pterygium Syndrome	AR	100	36 of 36
CHST14	Ehlers-Danlos Syndrome	AR	97.7	21 of 22
CHST3	Multiple Joint Dislocations, Craniofacial Dysmorphism, Spondyloepiphyseal Dysplasia, Skeletal Dysplasia	AR	99.97	38 of 38
CHSY1	Temtamy Preaxial Brachydactyly Syndrome	AR	96.64	13 of 16
CILK1	Endocrine-Cerebroosteodysplasia, Epilepsy	AD,AR	100	-
CLPTM1	Cleft Lip, Ogden Syndrome, Vander Woude Syndrome	-	97	3 of 3
CLPTM1L	Cleft Lip, Germ Cell Cander, Testicular Cancer	-	100	1 of 1
COG1	Congenital Disorder Of Glycosylation	AR	99.91	3 of 3
COG5	Congenital Disorder Of Glycosylation	AR	100	19 of 19
COL11A1	Deafness, Marshall Syndrome, Stickler Syndrome, Rhizomelic Dysplasia, Fibrochondrogenesis	AD,AR	100	104 of 106
COL11A2	Deafness, Fibrochondrogenesis, Otospondylomegaepiphyseal Dysplasia, Stickler Syndrome	AD,AR	99.98	58 of 58
COL2A1	Achondrogenesis, Necrosis Of Femoral Head, Multiple Dysplasia Types, Legg-Calve-Perthes Disease, Osteoarthritis, Stickler Syndrome, Dysspondyloenchondromatosis	AD,MU	100	583 of 583
COL4A1	Angiopathy, Nephropathy, Aneurysms, Leukoencephalopathy, Hanac Syndrome, Walker-Warburg Syndrome	AD	99.99	173 of 173
COL9A1	Epiphyseal Dysplasia, Stickler Syndrome	AD,AR	99.98	8 of 8
COL9A2	Epiphyseal Dysplasia, Stickler Syndrome	AD,AR	100	16 of 16
COL9A3	Epiphyseal Dysplasia, Stickler Syndrome	AD	99.98	20 of 20
COLEC10	3mc Syndrome	AR	99.95	3 of 3
COLEC11	Carnevale Syndrome, 3mc Syndrome	AR	100	11 of 11
COMT	Panic Disorder, Schizophrenia, 22q11.2 Deletion Syndrome	AD	99.98	5 of 5

CPLANE1	Joubert Syndrome, Varadi-Papp Syndrome, Monomelic Amyotrophy, Orofaciodigital Syndrome	AR	-	-
CPLX1	Epileptic Encephalopathy, Wolf-Hirschhorn Syndrome	AD,AR	99.81	3 of 3
CREBBP	Menke-Hennekam Syndrome, Rubinstein-Taybi Syndrome	AD	100	318 of 318
CRISPLD2	Cleft Lip/Palate, Van Der Woude Syndrome,	-	99	-
CRKL	22q11.2 Microdeletion Syndrome	-	99.93	5 of 6
CRPPA	Limb Girdle Muscular Dystrophy, Dystroglycanopathy, Walker-Warburg Syndrome	AR	97.69	-
CRYZ	Septic Myocarditis	-	99.92	-
CSPP1	Joubert Syndrome, Meckel Syndrome	AR	98.32	29 of 30
CTBP1	Hypotonia, Developmental Delay, Wolf-Hirschhorn Syndrome	AD	98.45	1 of 1
CTCF	Mental Retardation, Developmental Delay, Microcephaly	AD	96.6	39 of 41
CTNND1	Blepharocheilodontic Syndrome	AD	99.89	17 of 17
CYP26C1	Focal Facial Dermal Dysplasia	AR	97.95	14 of 14
CYTB	Leber Optic Atrophy, Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, Histiocytoid Cardiomyopathy, Neuropathy	MI	98.8	-
DAB1	Spinocerebellar Ataxia	AD	99.98	-
DAG1	Limb Girdle Muscular Dystrophy, Dystroglycanopathy, Muscle-Eye-Brain Disease, Leucodystrophy, Walker-Warburg Syndrome	AR	99.98	9 of 9
DCC	Colorectal Cancer, Esophageal Cancer, Gaze Palsy, Mirror Movements, Horizontal Gaze Palsy, Progressive Scoliosis, Kallmann Syndrome	AD,AR	94	39 of 39
DDX3X	Intellectual Developmental Disorder, Hypotonia	X,XR,XD,G	99.03	-
DDX59	Orofaciodigital Syndrome	AR	99.45	4 of 6
DEAF1	Dyskinesia, Seizures, Intellectual Developmental Disorder, Epilepsy, Extrapyramidal Syndrome, Smith-Magenis Syndrome	AD,AR	93.55	42 of 42
DENND4B	Alkuraya-Kucinskas Syndrome, Boucher-Neuhauser Syndrome, Oliver-Mcfarlane Syndrome, Mitochondrial Complex I Deficiency	-	99.95	1 of 1
DGCR2	Velocardiofacial Syndrome	AD	99.94	3 of 3
DGCR6	Velocardiofacial Syndrome	AD	94.78	-
DGCR8	Velocardiofacial Syndrome	AD	99.98	2 of 2
DHCR24	Desmosterolosis	AR	100	10 of 10
DHCR7	Smith-Lemli-Opitz Syndrome	AR	100	217 of 217
DHODH	Postaxial Acrofacial Dysostosis	AR	99	20 of 20
DISP1	Multiple Holoprosencephaly Types	-	100	10 of 10
DLG1	Cleft Lip/Palate	-	99.96	15 of 15
DLL1	Neurodevelopmental Disorder, Brain Abnormalities, Multiple Holoprosencephaly Types	AD	99.83	15 of 15
DLL3	Spondylocostal Dysostosis	AR	96.03	26 of 28
DLX4	Cleft Lip/Palate	AD	98.69	2 of 2
DLX5	Split-Hand And Foot Malformation, Hearing Loss	AD,AR	99.98	8 of 8
DMXL2	Deafness, Epileptic Encephalopathy, Polyendocrine-Polyneuropathy Syndrome	AD,AR	99.83	19 of 23
DOK7	Akinesia Deformation, Myasthenia, Myasthenic Syndromes	AR	99.88	72 of 72
DONSON	Microcephaly, Micromelia, Short Stature, Limb Abnormalities	AR	98.14	26 of 27
DPF2	Coffin-Siris Syndrome	AD	99.99	10 of 10
DPH1	Developmental Delay, Short Stature, Dysmorphic Features, Sparse Hair, Craniofacial Dysplasia, Ectodermal Anomalies, Intellectual Disability	AR	100	8 of 8
DSE	Ehlers-Danlos Syndrome	AR	99.94	3 of 3
DUSP22	Variola Major, T-Cell Lymphoma	-	100	-

DUSP6	Hypogonadotropic Hypogonadism, Kallmann Syndrome	AD,AR	99.36	4 of 4
DVL1	Robinow Syndrome	AD	100	20 of 20
DVL3	Robinow Syndrome	AD	100	16 of 16
DYNC2H1	Short-Rib Thoracic Dysplasia, Jeune Syndrome, Polydactyly	AR,MU,D	99.78	214 of 221
DYNC2I1	Short-Rib Thoracic Dysplasia, Polydactyly, Jeune Syndrome	AR	97.76	14 of 14
DYNC2I2	Short-Rib Thoracic Dysplasia, Polydactyly, Jeune Syndrome	AR	99.54	23 of 23
DYRK1A	Mental Retardation, 21q22.13q22.2 Microdeletion	AD	99.85	78 of 81
EARS2	Oxidative Phosphorylation Deficiency	AR	98.8	31 of 31
EBP	Mend Syndrome, Chondrodysplasia Punctata	X,XR,XD,G	100	-
ECEL1	Arthrogryposis	AR	99.52	39 of 39
EDN1	Auriculocondylar Syndrome, Question Mark Ears	AD,AR	100	9 of 9
EDNRA	Mandibulofacial Dysostosis, Alopecia, Migraine	AD	99.67	3 of 4
EFNB1	Craniofrontonasal Syndrome	X,XD,G	100	-
EFTUD2	Mandibulofacial Dysostosis, Microcephaly	AD	99.87	100 of 100
EIF2S3	Mehmo Syndrome	X,XR,G	98.64	-
EIF4A3	Robin Sequence, Cleft Mandible	AR	100	1 of 1
EPG5	Immunodeficiency, Cleft Lip/Palate, Cataract, Hypopigmentation, Corpus Callosum, Vici Syndrome	AR	98.98	73 of 73
ERCC4	Fanconi Anemia, Xeroderma Pigmentosum, Xfe Progeroid Syndrome, Cockayne Syndrome	AR	99.68	69 of 72
ERCC5	Cerebrooculofacioskeletal Syndrome, Xeroderma Pigmentosum, Cofs Syndrome, Cockayne Syndrome	AR	99.94	58 of 58
ESCO2	Roberts Syndrome, Sc Phocomelia Syndrome	AR	99.69	32 of 32
ESRP2	Cleft Lip/Palate, Laurence-Moon Syndrome, Atrial Septal Defect	-	99.08	5 of 5
ESS2	Velocardiofacial Syndrome	AD	99.91	-
EVC	Ellis-Van Creveld Syndrome, Weyers Acrofacial Dysostosis	AD,AR	94.04	68 of 73
EVC2	Ellis-Van Creveld Syndrome, Weyers Acrofacial Dysostosis	AD,AR	99.98	75 of 75
EXOC6B	Spondyloepimetaphyseal Dysplasia, Joint Laxity	AR	99.99	2 of 3
EYA1	Branchiootic Syndrome, Branchiootorenal Syndrome, Otofaciocervical Syndrome	AD	100	197 of 199
FAM149B1	Joubert Syndrome, Orofaciodigital Syndrome	AR	99.94	2 of 2
FAM20C	Raine Syndrome, Osteosclerotic Bone Dysplasia	AR	97.8	29 of 29
FAM50A	Mental Retardation Syndrome	X,XR,G	99.88	-
FANCA	Fanconi Anemia	AR	95.17	497 of 502
FANCB	Fanconi Anemia, Vacterl, Hydrocephalus	X,XR,G	95.53	-
FANCC	Fanconi Anemia	AR	100	75 of 75
FANCD2	Fanconi Anemia	AR	100	62 of 63
FANCE	Fanconi Anemia	AR	97	17 of 18
FANCF	Fanconi Anemia	AR	99.31	17 of 18
FANCG	Fanconi Anemia	-	100	94 of 94
FANCI	Fanconi Anemia	AR	100	53 of 54
FANCL	Fanconi Anemia	AR	100	25 of 26
FANCM	Spermatogenic Failure, Fanconi Anemia	AR	99.73	59 of 61
FBLN1	Synpolydactyly, Developmental Delay, Central Nervous System Anomaly	AD	98.03	4 of 4
FEZF1	Hypogonadotropic Hypogonadism, Kallmann Syndrome	AR	99.95	3 of 3
FGD1	Aarskog-Scott Syndrome	X,XR,G	98.95	-

FGF10	Aplasia Of Lacrimal And Salivary Glands, Lacrimoauriculodentodigital Syndrome,	AD	100	13 of 14
FGF17	Hypogonadotropic Hypogonadism, Kallmann Syndrome	AD,AR	99.98	8 of 8
FGF20	Renal Hypodysplasia, Renal Agenesis	AR	99.76	2 of 2
FGF8	Hypogonadotropic Hypogonadism, Holoprosencephaly, Kallmann Syndrome	AD	98.36	38 of 38
FGF9	Multiple Synostoses Syndrome	AD	100	2 of 2
FGFR1	Encephalocraniocutaneous Lipomatosis, Hartsfield Syndrome, Jackson-Weiss Syndrome, Kallmann Syndrome, Osteoglophonic Dysplasia, Pfeiffer Syndrome, Trigonocephaly, Holoprosencephaly, Hypogonadotropic Hypogonadism, Oligodontia	AD	100	279 of 280
FGFR2	Antley-Bixler Syndrome, Genital Anomalies, Apert Syndrome, Bent Bone Dysplasia Syndrome, Crouzon Syndrome, Cutis Gyrata Syndrome Of Beare And Stevenson, Scaphocephaly Syndrome, Jackson-Weiss Syndrome, Lacrimoauriculodentodigital Syndrome, Pfeiffer Syndrome, Saethre-Chotzen Syndrome	AD	98	140 of 143
FGFR3	Achondroplasia, Camptodactyly, Multiple Cancer Types, Crouzon Syndrome, Epidermal Nevus, Hypochondroplasia, Lacrimoauriculodentodigital Syndrome, Muenke Syndrome, Thanatophoric Dysplasia, Brachycephaly, Plagiocephaly, Saethre-Chotzen Syndrome	AD,AR	99.89	77 of 78
FGFRL1	Wolf-Hirschhorn Syndrome	AD	99.94	1 of 1
FKBP14	Ehlers-Danlos Syndrome, Kyphoscoliosis, Myopathy	AR	99.98	7 of 8
FKRP	Limb Girdle Muscular Dystrophy, Dystroglycanopathy, Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	99.9	157 of 157
FKTN	Cardiomyopathy, Limb Girdle Muscular Dystrophy, Dystroglycanopathy, Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	98	54 of 56
FLII	Smith-Magenis Syndrome	-	99.98	3 of 3
FLNA	Cardiac Valvular Dysplasia, Fg Syndrome, Frontometaphyseal Dysplasia, Heterotopia, Intestinal Pseudoobstruction, Melnick-Needles Syndrome, Otopalatodigital Syndrome, Terminal Osseous Dysplasia, Congenital Short Bowel Syndrome, Ehlers-Danlos Syndrome	X,XR,XD,G	100	-
FLNB	Atelosteogenesis, Boomerang Dysplasia, Larsen Syndrome, Spondylocarpotarsal Synostosis Syndrome	AD,AR	100	124 of 124
FLRT3	Hypogonadotropic Hypogonadism, Kallmann Syndrome	AD	99.98	7 of 7
FOXA2	Multiple Pituitary Hormone Deficiencies	-	97.66	7 of 8
FOXC2	Lymphedema-Distichiasis Syndrome	AD	87.59	54 of 95
FOXE1	Hypothyroidism, Cleft Palate, Thyroid Cancer, Athyreosis, Bamforth-Lazarus Syndrome, Papillary Or Follicular Thyroid Carcinoma	AD,AR	84.65	14 of 22
FOXF1	Alveolar Capillary Dysplasia	AD	95.93	74 of 96
FOXF2	Anterior Segment Dysgenesis, Cleft Lip/Palate, Axenfeld-Rieger Syndrome	-	85.21	3 of 4
FOXH1	Holoprosencephaly	-	98.72	30 of 33
FOXP2	Apraxia Of Speech	AD	100	17 of 17
FRAS1	Fraser Syndrome	AR	98.73	57 of 58
FREM2	Cryptophthalmos, Fraser Syndrome	AR	99.92	31 of 33
FTO	Growth Retardation, Developmental Delay	AR	99.91	8 of 8
FZD2	Omodysplasia, Robinow Syndrome	AD	98.63	7 of 7
FZD6	Nail Disorder	AR	99.98	16 of 16
G6PC3	Neutropenia	AR	100	45 of 45
GAS1	Holoprosencephaly	-	95.93	6 of 6
GATA1	Anemia, Neutropenia, Down Syndrome, Thrombocytopenia, Hemolysis, Beta-Thalassemia, Blackfan-Diamond Anemia, Erythropoietic Porphyria	X,XR,G	99.93	-

GATA3	Hypoparathyroidism, Deafness, Renal Disease	AD	100	81 of 81
GDF3	Klippel-Feil Syndrome, Microphthalmia	AD,MU,D	99.1	10 of 10
GDF6	Klippel-Feil Syndrome, Leber Congenital Amaurosis, Microphthalmia	AD,AR,MU,D	98.58	19 of 19
GJA1	Alopecia, Keratosis Palmoplantaris, Atrioventricular Septal Defect, Oculodentodigital Dysplasia, Craniometaphyseal Dysplasia, Erythrokeratodermia Variabilis, Left Heart Syndrome, Syndactyly	AD,AR,MU,O	100	119 of 119
GJB2	Deafness, Keratoderma, Knuckle Pads, Leukonychia, Kid Syndrome, Hyperkeratosis	AD,AR,X,XR,MU,D,G	99.89	413 of 419
GLI2	Holoprosencephaly, Pallister-Hall Syndrome, Pituitary Hormone Deficiencies	AD	98.38	83 of 88
GLI3	Greig Cephalopolysyndactyly Syndrome, Hypothalamic Hamartoma Syndrome, Pallister-Hall Syndrome, Polydactyly, Acrocallosal Syndrome, Tibial Hemimelia	AD,AR	100	231 of 231
GMNN	Meier-Gorlin Syndrome	AD	99.72	3 of 3
GMPPB	Limb Girdle Muscular Dystrophy, Dystroglycanopathy, Myasthenic Syndromes, Glycosylation Defect, Muscle-Eye-Brain Disease	AR	99.95	53 of 53
GNAI3	Auriculocondylar Syndrome	AD	99.95	12 of 12
GNAO1	Epileptic Encephalopathy, Neurodevelopmental Disorder	AD	100	47 of 47
GNB1	Leukemia, Mental Retardation, Developmental Delay, Seizures	AD,MU,P	100	31 of 31
GNRH1	Hypogonadotropic Hypogonadism	AR	100	12 of 12
GNRHR	Hypogonadotropic Hypogonadism	AR	100	59 of 59
GP1BB	Bernard-Soulier Syndrome, 22q11.2 Deletion Syndrome, Thrombocytopenia	AR	74.08	26 of 50
GPC3	Simpson-Golabi-Behmel Syndrome, Wilms Tumor, Nephroblastoma	AD,X,XR,G	99.84	-
GPC4	Keipert Syndrome, Simpson-Golabi-Behmel Syndrome, Wilms Tumor	AD,X,XR,G	98.43	-
GPC6	Omodysplasia	AR	99.92	3 of 3
GREB1L	Renal Agenesis	AD	97.94	41 of 41
GREM1	Hereditary Mixed Polyposis Syndrome	-	99.89	5 of 5
GRHL3	Van Der Woude Syndrome, Bifid Uvula, Cleft Velum	AD	99.99	29 of 29
GRIP1	Fraser Syndrome	AR	100	17 of 17
HAAO	Vertebral, Cardiac, Renal, And Limb Defects Syndrome	AR	100	2 of 2
HDAC8	Cornelia De Lange, Wilson-Turner Syndrome	X,XD,G	99.78	-
HES7	Spondylocostal Dysostosis	AR	85.76	4 of 6
HESX1	Septooptic Dysplasia, Pituitary Hormone Deficiencies, Hypothyroidism, Kallmann Syndrome, Pituitary Stalk Interruption Syndrome	AD,AR	100	26 of 26
HIRA	22q11.2 Deletion Syndrome	-	99.99	5 of 5
HNRNPK	Au-Kline Syndrome	AD	99.88	16 of 17
HOXA2	Microtia, Cleft Palate	AD,AR	99.93	5 of 5
HOXD13	Brachydactyly, Syndactyly, Synpolydactyly, Vacterl	AD	90.98	21 of 31
HS6ST1	Hypogonadotropic Hypogonadism, Kallmann Syndrome	AD	99.97	8 of 8
HSPG2	Dyssegmental Dysplasia, Schwartz-Jampel Syndrome	AR	99.41	68 of 69
HYAL1	Mucopolysaccharidosis, Hyaluronidase Deficiency	AR	99.83	4 of 4
HYAL2	Cor Triatriatum, Mucopolysaccharidosis, Cleft Lip/Palate	-	99.98	2 of 2
HYLS1	Hydrolethalus Syndrome, Joubert Syndrome	AR	100	2 of 2
IFT140	Retinitis Pigmentosa, Short-Rib Thoracic Dysplasia, Polydactyly, Jeune Syndrome, Leber Congenital Amaurosis	AR	99.97	81 of 81
IFT57	Orofaciodigital Syndrome	AR	99.8	2 of 2
IFT80	Asphyxiating Thoracic Dystrophy, Jeune Syndrome, Short Rib-Polydactyly Syndrome	AR	99.96	16 of 16

IFT88	Retinitis Pigmentosa	-	99.46	6 of 6
IGBP1	Corpus Callosum, Ocular Coloboma, Micrognathia	X,XR,G	100	-
IGF1R	Growth Delay, Insulin-Like Growth Factor I Resistance	AD,AR	100	72 of 73
IL17RD	Hypogonadotropic Hypogonadism, Kallmann Syndrome	AD,AR	99.95	17 of 17
IL1B	Gastric Cancer	AD	100	2 of 2
IL1RN	Gastric Cancer, Osteomyelitis, Periostitis, Pustulosis	AD,AR	100	10 of 11
INPP5E	Joubert Syndrome, Mental Retardation, Retinal Dystrophy	AR	99.89	56 of 56
INPPL1	Opsismodysplasia, Schneckenbecken Dysplasia	AR	95.41	29 of 31
INTS1	Neurodevelopmental Disorder, Cataracts, Poor Growth, Dysmorphic Facies	AR	98.94	9 of 9
INTU	Orofaciodigital Syndrome, Short-Rib Throacic Dysplasia, Polydactyly	AR	99.7	9 of 9
IQSEC2	Mental Retardation, Microduplication Xp11.22p11.23 Syndrome, Intellectual Disability, Microcephaly, Smith-Magenis Syndrome	X,XR,XD,G	99.73	-
IRF6	Orofacial Cleft, Popliteal Pterygium Syndrome, Van Der Woude Syndrome, Popliteal Pterygium Syndrome, Cleft Lip/Palate, Oligodontia	AD,MU,P	99.98	325 of 335
ISM1	Systemic Mastocytosis	-	99.17	-
ITGA8	Renal Hypodysplasia/Aplasia, Renal Agenesis	AR	99.68	7 of 7
JAG2	Alagille Syndorme, Hajdu-Cheney Syndrome, Cereblar Arteriopathy, Tongue Carcinoma	-	98.34	3 of 3
JMJD1C	22q11.2 Deletion Syndrome	-	99.09	27 of 27
JUP	Arrhythmogenic Right Ventricular Dysplasia, Naxos Disease, Acantholytic Epidermolysis Bullosa	AD,AR	100	56 of 56
KANSL1	Koolen-De Vries Syndrome	AD	96.03	22 of 27
KAT6A	Arboleda-Tham Syndrome, Intellectual Disability, Craniofacial Anomalies, Cardiac Defects Syndrome	AD	99.89	66 of 68
KAT6B	Genitopatellar Syndrome, Ohdo Syndrome, Blepharophimosis-Intellectual Disability Syndrome	AD	99.97	80 of 80
KATNIP	Joubert Syndrome	AR	99.97	7 of 7
KCNA1	Episodic Ataxia, Epileptic Encephalopathy, Continuous Muscle Fiber Activity, Dyskinesia	AD	100	49 of 49
KCNH1	Temple-Baraitser Syndrome, Zimmermann-Laband Syndrome	AD	99.69	15 of 15
KCNJ2	Andersen Cardiodysrhythmic Paralysis, Atrial Fibrillation, Short Qt Syndrome	AD	100	93 of 93
KCNK9	Birk-Barel Syndrome, Intellectual Disability	AD	100	3 of 3
KCNN3	Zimmermann-Laband Syndrome	AD	100	5 of 5
KDM1A	Cleft Palate, Developmental Delay	AD	98.18	16 of 16
KDM6A	Kabuki Syndrome	AD,X,XD,G	99.98	-
KIAA0586	Joubert Syndrome, Short-Rib Thoracic Dysplasia, Polydactyly	AR	99.84	31 of 32
KIAA0753	Orofaciodigital Syndrome	AR	97.73	7 of 7
KIF14	Meckel Syndrome, Microcephaly	AR	99.84	18 of 18
KIF7	Acrocallosal Syndrome, Hydrolethalus Syndrome, Macrocephaly, Epiphyseal Dysplasia, Orofaciodigital Syndrome	AR	94.91	47 of 50
KIFBP	Goldberg-Shprintzen Syndrome	AR	99.27	-
KISS1	Hypogonadotropic Hypogonadism	AR	100	9 of 10
KISS1R	Hypogonadotropic Hypogonadism, Precocious Puberty, Kallmann Syndrome	AD,AR	99.41	42 of 43
KLF4	Skin Squamous Cell Carcinoma, Epilepsy, Cerebral Cavernous Malformations	-	99.91	5 of 5
KLHL41	Nemaline Myopathy	AR	99.92	8 of 8
KLHL7	Crisponi/Cold-Induced Sweating Syndrome, Retinitis Pigmentosa, Bohring-Opitz Syndrome	AD,AR	98.69	19 of 19
KMT2A	Facial Dysmorphism, Developmental Delay, Cornelia De Lange, Wiedemann-Steiner Syndrome	AD	98.14	144 of 149

KMT2C	Kleefstra Syndrome	AD	98.76	55 of 59
KMT2D	Kabuki Syndrome	AD	99.71	839 of 847
KRAS	Aplasia Cutis Congenita, Arteriovenous Malformation Of The Brain, Cardiofaciocutaneous Syndrome, Leukemia, Noonan Syndrome, Autoimmune Lymphoproliferative Syndrome, Schimmelpenning-Feuerstein-Mims Syndrome, Encephalocraniocutaneous Lipomatosis, Nevus Sebaceus Syndrome, Lynch Syndrome, Toriello-Lacassie-Droste Syndrome	AD	100	38 of 38
LARGE1	Limb Girdle Muscular Dystrophy, Dystroglycanopathy, Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	100	-
LARS2	Hydrops Fetalis, Lactic Acidosis, Sideroblastic Anemia, Perrault Syndrome	AR	99.99	20 of 20
LBR	Pelger-Huet Anomaly, Reynolds Syndrome, Greenberg Dysplasia	AD,AR	99.98	34 of 34
LETM1	Wolf-Hirschhorn Syndrome	AD	98.2	2 of 2
LFNG	Spondylocostal Dysostosis	AR	84.38	9 of 11
LHX4	Pituitary Hormone Deficiency, Hypothyroidism, Pituitary Stalk Interruption Syndrome	AD	99.95	21 of 22
LHX8	Cleft Palate, Multiple Lipoma Types	-	100	2 of 2
LIG4	Lig4 Syndrome, Multiple Myeloma, Dubowitz Syndrome, Lig4 Syndrome, Omenn Syndrome	AR	99.48	46 of 46
LMNA	Cardiomyopathy, Charcot-Marie-Tooth Disease, Emery-Dreifuss Muscular Dystrophy, Heart-Hand Syndrome, Hutchinson-Gilford Progeria Syndrome, Malouf Syndrome, Mandibuloacral Dysplasia, Dermopathy, Werner Syndrome, Lipodystrophic Laminopathy, Hypergonadotropic Hypogonadism	AD,AR	100	619 of 620
LMX1B	9q33.3q34.11 Microdeletion Syndrome, Nail-Patella Syndrome	AD	100	191 of 191
LOXL3	Stickler Syndrome	-	99.97	7 of 7
LRP8	Myocardial Infarction, Lissencephaly, Cerebellar Hypoplasia	-	99.41	2 of 2
MAD2L2	Fanconi Anemia	AR	99.91	1 of 1
MAF	Ayme-Gripp Syndrome, Cataract	AD	75.14	23 of 23
MAFB	Duane Retraction Syndrome, Deafness, Osteolysis, Nephropathy	AD	98.63	24 of 24
MAP2K1	Cardiofaciocutaneous Syndrome, Melorheostosis, Noonan Syndrome	AD	100	31 of 31
MAP2K2	Cardiofaciocutaneous Syndrome, Neurofibromatosis, Noonan Syndrome	AD	100	37 of 37
MAP3K7	Cardiospondylocarpofacial Syndrome, Frontometaphyseal Dysplasia	AD	99.96	13 of 13
MAPK1	22q11.2 Microdeletion Syndrome	-	96.91	1 of 1
MAPRE2	Skin Creases	AD	99.98	5 of 5
MASP1	3mc Syndrome	AR	100	29 of 30
MBTPS2	Ichthyosis Follicularis, Photophobia Syndrome, Keratosis Follicularis Spinulosa Decalvans, Osteogenesis Imperfecta, Palmoplantar Keratoderma, Bresek Syndrome	X,XR,G	100	-
MCTP2	Monosomy 15q	-	99.95	6 of 6
MED12	Lujan-Fryns Syndrome, Ohdo Syndrome, Opitz-Kaveggia Syndrome, Blepharophimosis, Intellectual Disability, Fg Syndrome	X,XR,G	100	-
MED25	Basel-Vanagaite-Smirin-Yosef Syndrome, Charcot-Marie-Tooth Disease, Intellectual Disability, Cataract, Microcephaly, Nevus Flammeus Simplex	AR	100	5 of 5
MEGF10	Myopathy, Respiratory Distress, Dysphagia	AR	99.96	20 of 21
MEIS2	Cleft Palate, And Mental Retardation, 15q14 Microdeletion Syndrome	AD	92	18 of 20
MEOX1	Klippel-Feil Syndrome	AR	99.52	4 of 4
MESP2	Spondylocostal Dysostosis	AR	99.69	6 of 6
MIB1	Left Ventricular Noncompaction	AD	99.98	31 of 31
MID1	Opitz Syndrome	X,XR,G	99.95	-

MIR140	Spondyloepiphyseal Dysplasia	AD	-	-
MKKS	Bardet-Biedl Syndrome, Mckusick-Kaufman Syndrome	AR	89.96	71 of 71
MKS1	Bardet-Biedl Syndrome, Joubert Syndrome, Meckel Syndrome	AR	99.98	49 of 49
MN1	Cebalid Syndrome, Meningioma	AD	99.52	3 of 3
MSX1	Tooth Agenesis, Witkop Syndrome, Cleft Lip/Palate, Hypodontia, Dysplasia Of Nails Syndrome, Oligodontia	AD	99.54	50 of 51
MSX2	Craniosynostosis, Parietal Foramina, Cleidocranial Dysplasia	AD	99.98	15 of 15
MTHFR	Homocystinuria, Neural Tube Defects, Schizophrenia, Thrombophiliavenous Thromboembolism, Anencephaly, Exencephaly	AD,AR	100	122 of 122
MUSK	Akinesia Deformation Sequence, Myasthenic Syndrome	AR	95.58	23 of 25
MYH3	Arthrogryposis, Contractures, Spondylocarpostarsal Fusion Syndrome, Multiple Pterygium Syndrome, Digitotalar Dysmorphism, Freeman-Sheldon Syndrome, Sheldon-Hall Syndrome	AD,AR	100	46 of 47
MYMK	Carey-Fineman-Ziter Syndrome	AR	100	-
MYOD1	Myopathy, Diaphragmatic Defects, Respiratory Insufficiency, Dysmorphic Facies, Skinesia Deformation Sequence	AR	99.97	6 of 6
NAA10	Microphthalmia, Ogden Syndrome	X,XR,XD,G	99.86	-
NBAS	Liver Failure Syndrome, Short Stature, Optic Nerve Atrophy, Pelger-Huet Anomaly	AR	99.98	60 of 61
NBN	Aplastic Anemia, Leukemia, Nijmegen Breakage Syndrome, Breast And Ovarian Cancer	AR,MU,P	100	200 of 200
NDNF	Hypogonadotropic Hypogonadism, Kallmann Syndrome	AD	99.33	-
NEB	Nemaline Myopathy	AR	86.77	304 of 339
NECTIN1	Cleft Lip/Palate, Ectodermal Dysplasia	AR	100	-
NEDD4L	Periventricular Nodular Heterotopia	AD	97.61	10 of 10
NEK1	Amyotrophic Lateral Sclerosis, Short Rib-Polydactyly Syndrome, Orofaciodigital Syndrome	AD,AR,MU,D	99.83	73 of 74
NELFA	Wolf-Hirschhorn Syndrome	-	99.93	-
NEUROD2	Epileptic Encephalopathy	AD	96.88	2 of 2
NFASC	Neurodevelopmental Disorder, Motor Dysfunction	AR	100	12 of 12
NFIX	Marshall-Smith Syndrome, Sotos Syndrome, 19p13.3 Microduplication Syndrome, Malan Overgrowth Syndrome	AD	94.42	75 of 81
NID1	Isolated Dandy-Walker Malformation	-	100	4 of 4
NIPBL	Cornelia De Lange Syndrome	AD	99.32	409 of 426
NODAL	Heterotaxy, Holoprosencephaly	AD	100	18 of 18
NOTCH2	Acroosteolysis, Osteoporosis, Alagille Syndrome	AD	99.88	91 of 91
NPHP1	Joubert Syndrome, Nephronophthisis, Senior-Loken Syndrome, Bardet-Biedl Syndrome	AR	100	58 of 59
NSD2	Wolf-Hirschhorn Syndrome	AD	99.91	-
NSDHL	Ck Syndrome, Congenital Hemidysplasia, Ichthyosiform Erythroderma	X,XR,XD,G	100	-
NSMF	Hypogonadotropic Hypogonadism, Kallmann Syndrome	AD	99.69	11 of 11
NSUN2	Mental Retardation, Dubowitz Syndrome	AR	99.99	8 of 8
NTN1	Mirror Movements	AD	96.78	5 of 5
NUDT6	Gingival Overgrowth, Prostatic Adenoma, Desmoid Disease, Cardiac Valvular Dysplasia	-	99.87	1 of 1
NUP107	Galloway-Mowat Syndrome, Nephrotic Syndrome, Ovarian Dysgenesis, Gonadal Dysgenesis	AR	99.91	15 of 15
NUP88	Akinesia Deformation Sequence	AR	95.82	3 of 3
OFD1	Joubert Syndrome, Orofaciodigital Syndrome, Retinitis Pigmentosa, Simpson-Golabi-Behmel Syndrome, Orofaciodigital Syndrome, Primary Ciliary Dyskinesia	X,XR,XD,G	98.09	-

ORC1	Ear-Patella-Short Stature Syndrome	AR	100	12 of 12
ORC4	Meier-Gorlin Syndrome, Ear-Patella-Short Stature Syndrome	AR	100	4 of 4
ORC6	Meier-Gorlin Syndrome	AR	100	6 of 6
OTX2	Microphthalmia, Retinal Dystrophy, Pituitary Dysfunction, Agnathia-Holoprosencephaly, Situs Inversus Syndrome, Septo-Optic Dysplasia Spectrum	AD	100	56 of 58
PALB2	Fanconi Anemia, Familial Pancreatic Carcinoma, Breast And Ovarian Cancer	AD,AR	98.78	601 of 617
PAX3	Craniofacial-Deafness-Hand Syndrome, Rhabdomyosarcoma, Waardenburg Syndrome	AD,AR	99.98	157 of 157
PAX7	Myopathy, Scoliosis, Rhabdomyosarcoma	AR	100	17 of 17
PAX9	Tooth Agenesis, Selective, 3; Sthag3, Oligodontia	AD	100	53 of 55
PDE6D	Joubert Syndrome, Orofaciodigital Syndrome	AR	100	2 of 2
PDGFC	Milker's Nodule, Macular Degeneration, Medulloblastoma, Cleft Lip/Palate, Dermatitis	-	99.4	-
PDGFRA	Gastrointestinal Stromal Tumor, Gist-Plus Syndrome, Hypereosinophilic Syndrome	AD	100	24 of 24
PEX2	Refsum Disease, Neonatal Adrenoleukodystrophy, Zellweger Syndrome	AR	99.89	17 of 17
PEX5	Adrenoleukodystrophy, Cerebrohepatorenal Syndrome, Rhizomelic Chondrodysplasia Punctata, Refsum Disease, Zellweger Syndrome	AR	100	12 of 12
PEX7	Peroxisome Biogenesis Disorder, Refsum Disease, Rhizomelic Chondrodysplasia Punctata	AR	99.21	47 of 53
PGAP2	Hyperphosphatasia, Mental Retardation	AR	99.99	11 of 11
PGAP3	Hyperphosphatasia, Mental Retardation	AR	97	19 of 20
PGM1	Congenital Disorder Of Glycosylation	AR	99.96	38 of 40
PHF8	Intellectual Disability	X,XR,G	100	-
PHGDH	Neu-Laxova Syndrome, Phosphoglycerate Dehydrogenase Deficiency	AR	100	26 of 26
PHYH	Refsum Disease	AR	100	34 of 34
PIBF1	Joubert Syndrome	AR	99.83	7 of 7
PIEZO2	Arthrogryposis, Gordon Syndrome, Marden-Walker Syndrome	AD,AR	96.93	37 of 37
PIGA	Multiple Congenital Anomalies, Hypotonia, Seizures, Paroxysmal Nocturnal Hemoglobinuria, West Syndrome	X,XR,G	97.98	-
PIGL	Zunich Neuroectodermal Syndrome, Chime Syndrome, Hyperphosphatasia, Intellectual Disability	AR	86	11 of 13
PIGN	Hypotonia, Seizures, Fryns Syndrome	AR	93.97	36 of 39
PIGO	Hyperphosphatasia, Mental Retardation	AR	99.93	21 of 21
PIGP	Epileptic Encephalopathy	AR	99.98	2 of 2
PIGQ	Epileptic Encephalopathy	AR	99.99	4 of 4
PIGV	Hyperphosphatasia, Mental Retardation	AR	99.99	16 of 16
PIGW	Hyperphosphatasia, Mental Retardation	AR	99.52	6 of 6
PIGY	Hyperphosphatasia, Mental Retardation	AR	100	1 of 2
PLCB4	Auriculocondylar Syndrome	AD,AR	100	16 of 17
PLEKHA5	Cleft Lip/Palate, Blepharocheilodontic Syndrome	-	99.84	6 of 6
PLEKHA7	Renal Adenoma, Blepharocheilodontic Syndrome, Marshall Syndrome, Cleft Lip/Palate	-	100	7 of 7
PLXND1	Moebius Syndrome	-	98.44	6 of 6
PNKP	Ataxia, Oculomotor Apraxia, Charcot-Marie-Tooth Disease, Epileptic Encephalopathy, Ataxia, Oculomotor Apraxia	AR	100	36 of 36
POLA1	Pigmentary Disorder, Systemic Manifestations, Van Esch-O'driscoll Syndrome, Intellectual Disability	X,XR,G	99.26	-
POLR1A	Acrofacial Dysostosis, Choanal Atresia, Hearing Loss, Cardiac Defects, Craniofacial Dysmorphism	AD	99.8	6 of 6

POLR1B	Treacher-Collins Syndrome	AD	99.89	-
POLR1C	Leukodystrophy, Mandibulofacial Dysostosis, Hypogonadotropic Hypogonadism, Treacher-Collins Syndrome	AR	99.99	35 of 35
POLR1D	Treacher Collins Syndrome	AD,AR	100	23 of 23
POLR2A	Neurodevelopmental Disorder, Hypotonia, Intellectual And Behavioral Abnormalities,	AD	100	17 of 17
POLR3A	Leukodystrophy, Oligodontia, Hypogonadotropic Hypogonadism, Progeroid Syndrome, Wiedemann-Rautenstrauch Syndrome	AR	100	122 of 122
POMGNT1	Limb Girdle Muscular Dystrophy, Dystroglycanopathy, Retinitis Pigmentosa, Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	99.91	82 of 83
POMGNT2	Limb Girdle Muscular Dystrophy, Dystroglycanopathy, Walker-Warburg Syndrome	AR	100	10 of 10
POMK	Limb Girdle Muscular Dystrophy, Dystroglycanopathy, Walker-Warburg Syndrome	AR	99.99	8 of 8
POMT1	Limb Girdle Muscular Dystrophy, Dystroglycanopathy, Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	100	105 of 105
POMT2	Limb Girdle Muscular Dystrophy, Dystroglycanopathy, Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	100	74 of 74
PORCN	Dermal Hypoplasia	X,XD,G	100	-
POU1F1	Pituitary Hormone Deficiency, Hypothyroidism	AD,AR	100	43 of 44
PPP1R21	Fissured Tongue	-	100	7 of 7
PPP2R5D	Mental Retardation, Macrocephaly, Hypotonia, Behavioral Abnormalities	AD	100	11 of 11
PPP3CA	Arthrogryposis, Cleft Palate, Craniosynostosis, Intellectual Development, Epileptic Encephalopathy	AD	99.98	16 of 16
PQBP1	Renpenning Syndrome, Hamel Cerebro-Palato-Cardiac Syndrome, Intellectual Disability	X,XR,G	99.99	-
PRDM5	Brittle Cornea Syndrome	AR	99.86	13 of 13
PREPL	Myasthenic Syndrome, 2p21 Microdeletion Syndrome, Hypotonia-Cystinuria Syndrome	AR	99.92	7 of 12
PRICKLE1	Epilepsy, Unverricht-Lundborg Disease	AR	98.41	23 of 23
PROK2	Hypogonadotropic Hypogonadism, Kallmann Syndrome	AD	100	20 of 20
PROKR2	Hypogonadotropic Hypogonadism, Kallmann Syndrome, Pituitary Stalk Interruption Syndrome, Septo-Optic Dysplasia Spectrum	AD	100	64 of 64
PROP1	Pituitary Dwarfism, Pituitary Hormone Deficiencies, Hypothyroidism, Panhypopituitarism	AR	100	35 of 36
PRRX1	Agnathia-Holoprosencephaly-Situs Inversus Syndrome	AD,AR	100	11 of 11
PSAT1	Neu-Laxova Syndrome, Phosphoserine Aminotransferase Deficiency	AR	99.95	9 of 9
PTCH1	Basal Cell Carcinoma, Basal Cell Nevus Syndrome, Holoprosencephaly, Gorlin Syndrome, Monosomy 9q22.3	AD	98.89	498 of 502
PTCH2	Basal Cell Nevus Syndrome, Medulloblastoma, Gorlin Syndrome	AD,AR	99.98	11 of 11
PTDSS1	Lenz-Majewski Hyperostotic Dwarfism	AD	100	7 of 7
PTHLH	Brachydactyly	AD	100	12 of 12
PTPN11	Myelomonocytic Leukemia, Leopard Syndrome, Metachondromatosis, Noonan Syndrome	AD	100	150 of 151
PTPRS	Nk-Cell Enteropathy, Pituitary Hypoplasia, Pineal Region Meningioma	-	99.14	6 of 6
RAD21	Cornelia De Lange, Mungan Syndrome	AD,AR	99.8	16 of 17
RAD51	Fanconi Anemia, Mirror Movements, Breast And Ovarian Cancer	AD	99.98	16 of 16
RAD51C	Breast And Ovarian Cancer, Fanconi Anemia	AR	100	130 of 130
RAI1	17p11.2 Microduplication Syndrome, Smith-Magenis Syndrome	AD	99.91	50 of 53
RAPSN	Akinesia Deformation Sequence, Myasthenic Syndrome	AR	99.98	59 of 61
RB1	Bladder Cancer, Osteosarcoma, Retinoblastoma, Small Cell Cancer Of The Lung, Monosomy 13q14,	AD	99.41	941 of 995
RBM10	Tarp Syndrome	X,XR,G	100	-

RBM8A	Thrombocytopenia-Absent Radius Syndrome	AR	100	4 of 4
RECQL4	Baller-Gerold Syndrome, Rapadilino Syndrome, Rothmund-Thomson Syndrome	AR	96.72	134 of 135
RERE	Neurodevelopmental Disorder, 1p36 Deletion Syndrome	AD	92.43	21 of 21
RET	Multiple Endocrine Neoplasia, Thyroid Carcinoma, Haddad Syndrome, Pheochromocytoma, Paraganglioma, Hirschsprung Disease, Renal Agenesis	AD	100	453 of 454
REV3L	Moebius Syndrome	-	99.08	7 of 7
RFWD3	Fanconi Anemia	AR	99.99	2 of 2
RIC1	Catifa Syndrome	AR	99.9	-
RIMS3	Autism, Heimler Syndrome	-	100	2 of 2
RIPK4	Popliteal Pterygium Syndrome, Bartsocas-Papas Syndrome, Chand Syndrome	AR	99.98	16 of 16
RIPPLY2	Spondylocostal Dysostosis	AR	99.89	3 of 3
RNU4ATAC	Lowry-Wood Syndrome, Microcephalic Osteodysplastic Primordial Dwarfism, Roifman Syndrome	AR	-	-
RPGRIP1	Cone-Rod Dystrophy, Amaurosis, Meckel Syndrome	AR	99.33	146 of 159
RPGRIP1L	Coach Syndrome, Joubert Syndrome, Meckel Syndrome,	AR	99.96	52 of 52
RPL11	Blackfan-Diamond Anemia	AD	100	52 of 52
RPL15	Blackfan-Diamond Anemia	AD	99.74	8 of 9
RPL18	Blackfan-Diamond Anemia	AD	100	1 of 1
RPL26	Blackfan-Diamond Anemia	AD	92.97	1 of 1
RPL27	Blackfan-Diamond Anemia	AD	100	2 of 2
RPL31	Blackfan-Diamond Anemia	-	100	0 of 1
RPL35	Blackfan-Diamond Anemia	AD	100	1 of 1
RPL35A	Blackfan-Diamond Anemia	AD	100	12 of 12
RPL5	Blackfan-Diamond Anemia	AD	100	95 of 95
RPS10	Blackfan-Diamond Anemia	AD	100	7 of 7
RPS15A	Blackfan-Diamond Anemia	AD	98.74	1 of 1
RPS17	Blackfan-Diamond Anemia	AD	0	0 of 7
RPS19	Blackfan-Diamond Anemia	AD	78	159 of 165
RPS23	Macinnes Syndrome	AD	99.99	2 of 2
RPS24	Blackfan-Diamond Anemia	AD	90.17	11 of 14
RPS26	Blackfan-Diamond Anemia	AD	100	28 of 29
RPS27	Blackfan-Diamond Anemia	AD	99.85	1 of 1
RPS28	Diamond-Blackfan Anemia, Mandibulofacial Dysostosis, Blackfan-Diamond Anemia	AD	100	1 of 1
RPS29	Blackfan-Diamond Anemia	AD	100	4 of 4
RPS7	Blackfan-Diamond Anemia	AD	100	7 of 10
RREB1	22q11.2 Deletion Syndrome	-	99.92	8 of 8
RSPO2	Humerofemoral Hypoplasia, Radiotibial Ray Deficiency, Tetraamelia Syndrome	AR	99.67	-
RUNX2	Cleidocranial Dysplasia, Metaphyseal Dysplasia With Maxillary Hypoplasia And Brachydactyly, Cleidocranial Dysplasia	AD	73.67	189 of 190
RXYLT1	Limb Girdle Muscular Dystrophy, Dystroglycanopathy, Walker-Warburg Syndrome	AR	99.46	-
RYK	Mieloid Leukemia, Exudative Vitreoretinopathy, Robinow Syndrome, Norrie Disease	-	90.27	1 of 1
RYR1	Central Core Disease Of Muscle, Malignant Hyperthermia, Minicore Myopathy, Ophthalmoplegia	AD,AR	97.63	733 of 746
SALL4	Duane-Radial Ray Syndrome, Ivic Syndrome, Acro-Renal-Ocular Syndrome	AD	100	54 of 54

SATB2	2q32-Q33 Deletion Syndrome	AD	99.87	97 of 124
SCARF2	Van Den Ende-Gupta Syndrome	AR	93.06	13 of 13
SCN1B	Atrial Fibrillation, Brugada Syndrome, Epileptic Encephalopathy, Dravet Syndrome, Cardiac Conduction Defect	AD,AR	99.67	46 of 48
SCN2A	Epileptic Encephalopathy, Episodic Ataxia, Seizures, Dravet Syndrome, West Syndrome	AD	100	351 of 351
SEC23A	Craniolenticulosutural Dysplasia	AR	100	4 of 4
SEC24C	22q11.2 Deletion Syndrome	-	99.98	-
SELENOI	Spastic Paraplegia, Kennedy Pathway Dysfunction	AR	99.86	-
SEMA3A	Hypogonadotropic Hypogonadism, Brugada Syndrome, Kallmann Syndrome	AD	100	29 of 29
SEMA3E	Charge Syndrome, Hypogonadotropic Hypogonadismia	AD,AR	99.81	6 of 7
SEPTIN9	Amyotrophy, Neuralgic Amyotrophy	AD	86.94	4 of 4
SETD5	Mental Retardation, Cornelia De Lange, Intellectual Disability, Facial Dysmorphism,	AD	99.77	37 of 37
SF3B4	Nager Synd, Acrofacial Dysostosis	AD	94.86	33 of 40
SH2B1	16p11.2 Microdeletion Syndrome, Obesity, Insulin Resistance	-	99.98	25 of 25
SHH	Holoprosencephaly, Microphthalmia, Schizencephaly, Polydactyly, Radial Hemimelia, Syndactyly	AD	99.48	161 of 184
SIK1	Epileptic Encephalopathy, West Syndrome	AD	99.67	9 of 9
SIX1	Branchiootic Syndrome, Branchiootorenal Syndrome, Deafness, Bor Syndrome	AD	73	20 of 20
SIX3	Holoprosencephaly, Schizencephaly	AD	99.79	79 of 80
SIX5	Branchiootorenal Syndrome, Bor Syndrome	AD	93.16	11 of 11
SKI	1p36 Deletion Syndrome, Shprintzen-Goldberg Syndrome	AD	99.66	39 of 39
SLC10A7	Short Stature	AR	99.99	8 of 8
SLC18A3	Myasthenic Syndrome, Kinesia Deformation Sequence	AR	99.97	5 of 5
SLC25A19	Microcephaly, Thiamine Metabolism Dysfunction Syndrome	AR	97.13	10 of 10
SLC25A22	Epileptic Encephalopathy	AR	100	16 of 16
SLC26A2	Achondrogenesis, Atelosteogenesis, Diastrophic Dysplasia, Epiphyseal Dysplasia, Diastrophic Dwarfism	AR	99.59	51 of 56
SLC29A3	Histiocytosis-Lymphadenopathy Plus Syndrome, Dysosteosclerosis, H Syndrome	AR	100	32 of 32
SLC2A10	Arterial Tortuosity Syndrome	AR	100	35 of 35
SLC35D1	Schneckenbecken Dysplasia	AR	99.98	7 of 8
SLC39A13	Ehlers-Danlos Syndrome	AR	100	9 of 9
SLX4	Fanconi Anemia	AR	99.92	76 of 76
SMAD2	Osteopoikilosis, Buschke-Ollendorff Syndrome, Hypertrophic Scars, Clear Cell Adenocarcinoma, Melorheostosis	-	100	19 of 19
SMAD3	Loeys-Dietz Syndrome, Aneurysm-Osteoarthritis Syndrome	AD	100	128 of 128
SMAD4	Polyposis Syndrome, Myhre Syndrome, Pancreatic Cancer, Thoracic Aortic Aneurysm And Aortic Dissection, Hemorrhagic Telangiectasia	AD	99.56	136 of 136
SMARCA4	Coffin-Siris Syndrome, Rhabdoid Tumor Predisposition Syndrome	AD	100	68 of 69
SMARCB1	Coffin-Siris Syndrome, Rhabdoid Tumor Predisposition Syndrome, Schwannomatosis, Meningioma	AD	100	97 of 99
SMARCC2	Coffin-Siris Syndrome	AD	99.49	16 of 16
SMARCD1	Coffin-Siris Syndrome	AD	93.17	7 of 7
SMARCE1	Coffin-Siris Syndrome, Meningioma	AD	98.98	15 of 15
SMC1A	Cornelia De Lange, Holoprosencephaly, Wiedemann-Steiner Syndrome	X,XR,XD,G	100	-
SMC3	Cornelia De Lange Syndrome	AD	100	30 of 30

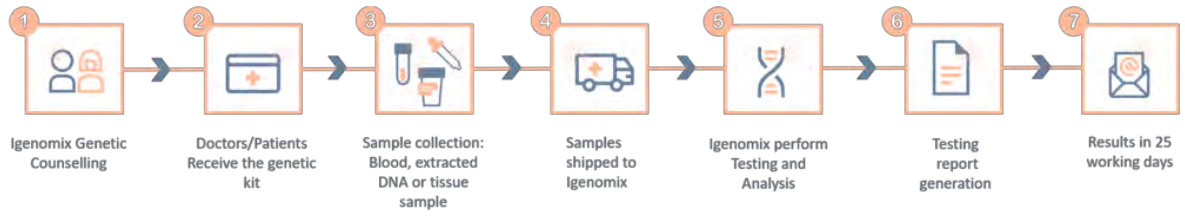
SMCHD1	Bosma Arhinia Microphthalmia Syndrome, Facioscapulohumeral Muscular Dystrophy, Hyposmia, Nasal And Ocular Hypoplasia, Hypogonadotropic Hypogonadism	AD,MU,D	99.64	131 of 137
SMG1	Myoclonus Epilepsy, Limb Girdle Muscular Distrophy, Sechel Syndrome	-	98.02	1 of 1
SMG9	Heart And Brain Malformation Syndrome	AR	100	4 of 4
SMO	Basal Cell Carcinoma, Craniofacial Malformations, Polysyndactyly, Abnormal Skin, Gut Development, Hypothalamic Hamartoma Syndrome, Curry-Jones Syndrome, Meningioma	AR	94.03	10 of 10
SMOC1	Microphthalmia, Limb Anomalies	AR	100	19 of 19
SMPD4	Neurodevelopmental Disorder, Microcephaly, Arthrogryposis	AR	99.98	15 of 15
SMS	Mental Retardation	X,XR,G	84.04	-
SNAP29	Cerebral Dysgenesis, Neuropathy, Ichthyosis, Palmoplantar Keratoderma, Cednik Syndrome	AR	100	13 of 13
SNRPB	Cerebrocostomandibular Syndrome	AD	98.97	-
SNTG1	Idiopahic Scoliosis, Basal Ganglia Calcification	-	100	1 of 2
SON	Zttk Syndrome, Brain Malformations, Musculoskeletal Abnormalities, Facial Dysmorphism, Intellectual Disability	AD	99.27	30 of 32
SOX10	Peripheral Demyelinating Neuropathy, Waardenburg Syndrome, Kallmann Syndrome, Central Dysmyelinating Leukodystrophy, Hirschsprung Disease	AD	99.74	139 of 147
SOX11	Mental Retardation, Coffin-Siris Syndrome	AD	95.23	11 of 11
SOX2	Microphthalmia, Anophthalmia, Esophageal Atresia Syndrome, Septo-Optic Dysplasia Spectrum	AD	99.91	78 of 78
SOX3	Mental Retardation, Growth Hormone Deficiency, Panhypopituitarism, Testicular Disorder Of Sex Development, Septo-Optic Dysplasia Spectrum	X,G	92.88	-
SOX4	Coffin-Siris Syndrome	AD	75.52	4 of 4
SOX6	Tolchin-Le Caignec Syndrome	AD	99.98	2 of 2
SOX9	Campomelic Dysplasia, Ovotesticular Disorder Of Sex Development, Gonadal Dysgenesis, Pierre Robin Syndrome	AD	97.28	87 of 95
SP8	Retinitis Pigmentosa, Bleeding Disorders	-	94.48	1 of 3
SPECC1L	Facial Clefting, Hypertelorism, Opitz Gbbb Syndrome	AD	99.66	14 of 14
SPEG	Myopathy	AR	99.26	17 of 17
SPOP	Nabais Sa-De Vries Syndrome	AD	100	1 of 1
SPRY2	Iga Nephropathy	AD	99.87	4 of 4
SPRY4	Hypogonadotropic Hypogonadism, Kallmann Syndrome	AD,AR	99.72	13 of 13
STAC3	Native American Myopathy	AR	99.98	5 of 5
STAG2	Holoprosencephaly, Neurodevelopmental Disorder, Craniofacial Abnormalities, Xq25 Microduplication Syndrome	X,XR,G	99.09	-
STAMBP	Microcephaly, Capillary Malformation Syndrome	AR	99.88	21 of 22
STAT3	Autoimmune Disease, Hyper-Ige Recurrent Infection Syndrome, Promyelocytic Leukemia, Diabetes Mellitus	AD	100	171 of 171
STXBP1	Epileptic Encephalopathy, 9q33.3q34.11 Microdeletion Syndrome, Atypical Rett Syndrome, Dravet Syndrome, West Syndrome	AD	100	209 of 215
SUFU	Basal Cell Nevus Syndrome, Joubert Syndrome, Medulloblastoma, Meningioma, Acrocallosal Syndrome, Gorlin Syndrome, Holoprosencephaly	AD,AR	99.99	43 of 43
SUMO1	Orofacial Cleft, Oligodontia	-	99.77	-
TAC3	Hypogonadotropic Hypogonadism	AR	100	10 of 10
TACR3	Hypogonadotropic Hypogonadism, Kallmann Syndrome	AR	99.97	40 of 40
TAPT1	Osteochondrodysplasia	AR	89.49	3 of 3
TBC1D24	Deafness, Doors Syndrome, Epileptic Encephalopathy, Paroxysmal Exercise, Writer's Cramp Syndrome	AD,AR	100	80 of 80
TBCE	Encephalopathy, Hypoparathyroidism, Dysmorphism, Kenny-Caffey Syndrome, Spastic Ataxia, Sanjad-Sakati Syndrome	AR	100	8 of 8

TBR1	Intellectual Developmental Disorder, Autism, Speech Delay, 2q24 Microdeletion Syndrome	AD	99.04	13 of 13
TBX1	Conotruncal Heart Malformations, Digeorge Syndrome, Tetralogy Of Fallot, Velocardiofacial Syndrome, 22q11.2 Deletion Syndrome, 22q11.2 Microduplication Syndrome	AD,AR	88.7	35 of 42
TBX10	Cleft Lip/Palate, Digeorge Syndrome, Immunodeficiency	-	100	1 of 1
TBX15	Pelviscapular Dysplasia	AR	100	3 of 3
TBX2	Vertebral Anomalies	AD	93.43	8 of 13
TBX22	Charge-Like Syndrome, Cleft Palate, Abruzzo-Erickson Syndrome	X,G	99.94	-
TBX4	Amelia, Ischiocoxopodopatellar Syndrome, 17q23.1q23.2 Microdeletion Syndrome, Coxopodopatellar Syndrome, Clubfoot	AD,AR	99.72	91 of 94
TBX6	Costovertebral Segmentation Anomalies, Spondylocostal Dysostosis	AD,AR	99.89	32 of 33
TCOF1	Treacher-Collins Syndrome	AD	100	326 of 327
TCTN2	Joubert Syndrome, Meckel Syndrome	AR	100	14 of 14
TCTN3	Joubert Syndrome, Orofaciodigital Syndrome	AR	99.99	13 of 13
TDGF1	Holoprosencephaly	-	100	3 of 3
TELO2	You-Hoover-Fong Syndrome, Intellectual Disability, Neurodevelopmental Disorder	AR	99.98	8 of 8
TFAP2A	Branchiooculofacial Syndrome	AD	98.61	37 of 37
TGDS	Catel-Manzke Syndrome	AR	99.99	7 of 7
TGFB1	Camurati-Engelmann Disease, Cystic Fibrosis, Inflammatory Bowel Disease, Immunodeficiency, Encephalopathy	AD,AR	99.75	24 of 24
TGFB2	Loeys-Dietz Syndrome, Aortic Aneurysm	AD	99.9	41 of 44
TGFB3	Arrhythmogenic Right Ventricular Dysplasia, Loeys-Dietz Syndrome, Aortic Aneurysm, Aortic Dissection	AD	100	34 of 35
TGFBR1	Loeys-Dietz Syndrome, Multiple Self-Healing Squamous Epithelioma, Aortic Aneurysm, Aortic Dissection	AD	94	96 of 100
TGFBR2	Colorectal Cancer, Esophageal Cancer, Loeys-Dietz Syndrome, Aortic Aneurysm, Aortic Dissection, Lynch Syndrome, Squamous Cell Carcinoma Of The Esophagus	AD	99.9	165 of 166
TGIF1	Holoprosencephaly	AD	99.94	23 of 23
TMCO1	Cerebrofaciothoracic Dysplasia	AR	88	5 of 5
TMEM107	Meckel Syndrome, Orofaciodigital Syndrome	AR	100	3 of 3
TMEM216	Joubert Syndrome, Meckel Syndrome, Orofaciodigital Syndrome	AR	98.74	8 of 8
TMEM231	Joubert Syndrome, Meckel Syndrome, Orofaciodigital Syndrome	AR	98.63	20 of 21
TMEM237	Joubert Syndrome	AR	100	11 of 11
TMEM67	Bardet-Biedl Syndrome, Coach Syndrome, Joubert Syndrome, Meckel Syndrome, Nephronophthisis, Rhyns Syndrome	AR	96.93	177 of 179
TP63	Adult Syndrome, Ankyloblepharon, Ectodermal Defects, Cleft Lip/Palate, Ectrodactyly, Limb-Mammary Syndrome, Rapp-Hodgkin Syndrome, Split-Hand/Foot Malformation, Bladder Exstrophy, Eec Syndrome	AD	99.98	144 of 144
TRAPPC9	Mental Retardation	AR	100	17 of 18
TRIM8	Epileptic Encephalopathy	-	99.5	7 of 7
TRIP13	Mosaic Variegated Aneuploidy Syndrome, Nephroblastoma	AR	98.14	2 of 2
TRPV4	Necrosis Of Femoral Head, Brachyrachia, Digital Arthropathy, Hereditary Motor, Sensory Neuropathy, Metatropic Dysplasia, Parastremmatic Dwarfism, Spinal Muscular Atrophy, Spondyloepiphyseal Dysplasia	AD	100	88 of 88
TRRAP	Deafness, Developmental Delay, Autism	AD	99.98	46 of 46
TSR2	Blackfan-Diamond Anemia	X,XR,G	99.96	-
TTC37	Trichohepatoenteric Syndrome, Syndromic Diarrhea	AR	100	66 of 66
TTN	Cardiomyopathy, Limb Girdle Muscular Dystrophy, Myopathy	AD,AR	97.93	1153 of 1219

TUBB	Cortical Dysplasia, Skin Creases	AD	100	8 of 8
TWIST1	Craniosynostosis, Robinow-Sorauf Syndrome, Saethre-Chotzen Syndrome, Sweeney-Cox Syndrome, Brachycephaly, Plagiocephaly, Scaphocephaly	AD	74.06	133 of 161
TXNL4A	Burn-Mckeown Syndrome, Choanal Atresia, Hearing Loss, Cardiac Defects, Craniofacial Dysmorphism	AR	80.96	4 of 4
TXNRD2	Glucocorticoid Deficiency, Cardiomyopathy	AR	99.95	5 of 5
UBB	Bifid Uvula, Cleft Velum	-	100	1 of 1
UBE2T	Fanconi Anemia	AR	100	4 of 4
UFD1	22q11.2 Deletion Syndrome	-	99.98	-
USP9X	Mental Retardation, Facial Dysmorphism, Short Stature, Choanal Atresia	X,XR,XD,G	98.61	-
VANGL2	Neural Tube Defects, Anencephaly, Exencephaly	AD	99.98	12 of 12
VAX1	Microphthalmia	AR	88.33	2 of 4
WASHC5	Dandy-Walker Malformation, Spastic Paraplegia	AD,AR	99.99	-
WDPCP	Bardet-Biedl Syndrome, Heart Defects, Tongue Hamartomas, Polysyndactyly, Meckel Syndrome	AR	99.3	8 of 8
WDR11	Hypogonadotropic Hypogonadism, Kallmann Syndrome, Pituitary Stalk Interruption Syndrome	AD,AR	100	19 of 19
WDR26	Skraban-Deardorff Syndrome, Intellectual Disability, Seizures, Abnormal Gait, Facial Dysmorphism	AD	99.31	22 of 22
WDR35	Cranioectodermal Dysplasia, Short-Rib Thoracic Dysplasia, Polydactyly	AR	100	31 of 33
WHCR	Wolf-Hirschhorn Syndrome	AD	-	-
WNT3	Tetra-Amelia	AR	100	2 of 2
WNT4	Sex Reversal, Kidneys Dysgenesis, Mullerian Aplasia And Hyperandrogenism, Serkal Syndrome	AD,AR	100	8 of 8
WNT5A	Robinow Syndrome	AD	99.08	11 of 11
WNT7A	Fibular Aplasia, Hypoplasia, Oligodactyly, Phocomelia	AR	100	10 of 10
XRCC2	Fanconi Anemia, Male Infertility, Azoospermia, Oligozoospermia	AR	98.39	28 of 28
XYLT1	Pseudoxanthoma Elasticum, Desbuquois Syndrome	AR	92.61	19 of 23
YAP1	Coloboma, Hearing Impairment, Cleft Lip/Palate, Cleft Lip/Palate	AD	99.88	8 of 8
YWHAE	17p13.3 Microduplication Syndrome, Miller-Dieker Syndrome	-	98.99	0 of 1
ZBTB24	Immunodeficiency, Centromeric Instability, Facial Anomalies Syndrome, Icf Syndrome	AR	100	23 of 23
ZC4H2	Wieacker-Wolff Syndrome, Intellectual Disability, Developmental Delay	X,XR,XD,G	99.69	-
ZEB2	Mowat-Wilson Syndrome	AD	98.95	253 of 254
ZIC2	Holoprosencephaly	AD	84.47	86 of 112
ZMPSTE24	Mandibuloacral Dysplasia, Lipodystrophy, Restrictive Dermopathy, Hutchinson-Gilford Progeria Syndrome	AR	100	35 of 36
ZNF469	Brittle Cornea Syndrome	AR	99.91	79 of 79
ZSWIM6	Acromelic Frontonasal Dysostosis, Neurodevelopmental Disorder, Movement Abnormalities, Autism	AD	91.16	2 of 2

*Inheritance: AD: Autosomal Dominant; AR: Autosomal Recessive; X: X linked; XLR: X linked Recessive; Mi: Mitochondrial; Mu: Multifactorial.
**Number of clinically relevant mutations according to HGMD

Methodology



Contact us

Call +34 963 905 310 or send an email to supportspain@igenomix.com for any of the following objectives:

- Get more information about the test.
- Request your kit.
- Request a pick up of the kit after collecting the sample.

References

1. Voigt, A., Radlanski, R. J., Sarioglu, N., & Schmidt, G. (2017). Lippen-Kiefer-Gaumen-Spalten [Cleft lip and palate]. *Der Pathologe*, *38*(4), 241–247. https://doi.org/10.1007/s00292-017-0313-x
2. Abualfaraj, R., Daly, B., McDonald, F., & Scambler, S. (2018). Cleft lip and palate in context: Learning from, and adding to, the sociological literature on long-term conditions. *Health (London, England : 1997)*, *22*(4), 372–388. https://doi.org/10.1177/1363459317693409
3. Farronato, G., Cannalire, P., Martinelli, G., Tubertini, I., Giannini, L., Galbiati, G., & Maspero, C. (2014). Cleft lip and/or palate: review. *Minerva stomatologica*, *63*(4), 111–126.
4. Worley, M. L., Patel, K. G., & Kilpatrick, L. A. (2018). Cleft Lip and Palate. *Clinics in perinatology*, *45*(4), 661–678. https://doi.org/10.1016/j.clp.2018.07.006
5. Mossey, P. A., Little, J., Munger, R. G., Dixon, M. J., & Shaw, W. C. (2009). Cleft lip and palate. *Lancet (London, England)*, *374*(9703), 1773–1785. https://doi.org/10.1016/S0140-6736(09)60695-4
6. Dental Supplement, Candotto, V., Oberti, L., Gabrione, F., Greco, G., Rossi, D., Romano, M., & Mummolo, S. (2019). Current concepts on cleft lip and palate etiology. *Journal of biological regulators and homeostatic agents*, *33*(3 Suppl. 1), 145–151.
7. Setó-Salvia, N., & Stanier, P. (2014). Genetics of cleft lip and/or cleft palate: association with other common anomalies. *European journal of medical genetics*, *57*(8), 381–393. https://doi.org/10.1016/j.ejmg.2014.04.003