

## Craniosynostosis

### Precision Panel



### Overview

Craniosynostosis is defined as the premature fusion of one or more cranial sutures, often resulting in abnormal head shape. It is a developmental craniofacial anomaly resulting from a primary defect of ossification (primary craniosynostosis) or, more commonly, from a failure of brain growth (secondary craniosynostosis). As well, craniosynostosis can be simple when only one suture fuses prematurely or complex/compound when there is a premature fusion of multiple sutures. Complex craniosynostosis are usually associated with other body deformities. The main morbidity risk is the elevated intracranial pressure and subsequent brain damage. When left untreated, craniosynostosis can cause serious complications such as developmental delay, facial abnormality, sensory, respiratory and neurological dysfunction, eye anomalies and psychosocial disturbances. In approximately 85% of the cases, this disease is isolated and nonsyndromic. Syndromic craniosynostosis usually present with multiorgan complications.

The Igenomix Craniosynostosis Precision Panel can be used to make a directed and accurate diagnosis 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 Craniosynostosis Precision Panel is indicated for those patients with a clinical diagnosis or suspicion with or without the following manifestations:

- Microcephaly
- Scaphocephaly (elongated head)
- Anterior plagiocephaly
- Brachycephaly
- Torticollis
- Frontal bossing

### 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 treatment in the form surgical procedures to relieve fused sutures, midface advancement, limited phase of orthodontic treatment and combined

orthodontics/orthognathic surgery treatment. Monitoring and prevent complications of elevated intracranial pressure.

- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.
- Improvement of delineation of genotype-phenotype correlation.

## Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
<b>ADAMTS3</b>	Hennekam Syndrome	AR	99.97	4 of 4
<b>AHDC1</b>	Xia-Gibbs Syndrome, AHDC1-Related Intellectual Disability-Obstructive Sleep Apnea-Mild Dysmorphism Syndrome	AD	99.87	41 of 43
<b>AKT1</b>	Proteus Syndrome, Meningioma	AD	100	6 of 6
<b>ALPL</b>	Hypophosphatasia	AD,AR	100	320 of 321
<b>ALX3</b>	Frontonasal Dysplasia	AR	89.31	8 of 8
<b>ALX4</b>	Craniosynostosis, Frontonasal Dysplasia, Enlarged Parietal Foramina, Isolated Scaphocephaly, Potocki-Shaffer Syndrome	AD,AR	99.94	25 of 25
<b>APC2</b>	Complex Cortical Dysplasia, Sotos Syndrome	AR	94.97	11 of 11
<b>ARVCF</b>	22q11.2 Deletion Syndrome	-	99.95	2 of 2
<b>ASXL1</b>	Bohring-Opitz Syndrome, Bohring-Opitz Syndrome	AD	99.96	41 of 41
<b>ATR</b>	Cutaneous Telangiectasia And Cancer Syndrome, Seckel Syndrome	AD,AR	99.98	39 of 40
<b>ATRIP</b>	Seckel Syndrome	-	99.89	2 of 2
<b>B3GAT3</b>	Multiple Joint Dislocations, Short Stature, Craniofacial Dysmorphism, With Or Without Congenital Heart Defects	AR	99.86	15 of 15
<b>B3GLCT</b>	Peters Plus Syndrome	AR	99.96	-
<b>B4GALT7</b>	Ehlers-Danlos Syndrome, Spondylodysplastic Type	AR	99.92	11 of 11
<b>BMP4</b>	Syndromic Microphthalmia, Cleft Lip/Palate, Microphthalmia With Brain And Digit Anomalies	AD,MU,P	100	38 of 42
<b>BPNT2</b>	Chondrodysplasia With Joint Dislocations	AR	97.04	4 of 4
<b>CCBE1</b>	Hennekam Lymphangiectasia-Lymphedema Syndrome	AR	100	16 of 16
<b>CCNQ</b>	Toe Syndactyly, Telecanthus, And Anogenital And Renal Malformations	X,XD,G	99.59	-
<b>CD96</b>	C Syndrome	AD	100	4 of 4
<b>CDC45</b>	Meier-Gorlin Syndrome, Ear-Patella-Short Stature Syndrome	AR	99.99	19 of 19
<b>CDC6</b>	Meier-Gorlin Syndrome, Ear-Patella-Short Stature Syndrome	AR	100	2 of 2
<b>CDH11</b>	Elsahy-Waters Syndrome, Branchioskeletogenital Syndrome	AR	99.95	10 of 10
<b>CDT1</b>	Meier-Gorlin Syndrome, Ear-Patella-Short Stature Syndrome	AR	97.43	12 of 12
<b>CENPE</b>	Primary Microcephaly, Seckel Syndrome	AR	95.69	5 of 5
<b>CENPJ</b>	Autosomal Recessive Primary Microcephaly, Seckel Syndrome	AR	99.97	13 of 13
<b>CEP120</b>	Joubert Syndrome, Short-Rib Thoracic Dysplasia With Or Without Polydactyly, Jeune Syndrome	AR	99.8	9 of 9
<b>CEP152</b>	Autosomal Recessive Primary Microcephaly, Seckel Syndrome	AR	97.73	21 of 24
<b>CHST14</b>	Musculocontractural Ehlers-Danlos Syndrome	AR	97.7	21 of 22
<b>CHST3</b>	Multiple Joint Dislocations, Short Stature, Craniofacial Dysmorphism, With Or Without Congenital Heart Defects, Spondyloepiphyseal Dysplasia With Congenital Joint Dislocations, Chst3-Related Skeletal Dysplasia	AR	99.97	38 of 38
<b>CLCN7</b>	Osteopetrosis, Albers-Schonberg Osteopetrosis	AD,AR	99.85	109 of 111
<b>COLEC10</b>	3mc Syndrome	AR	99.95	3 of 3
<b>COLEC11</b>	Carnevale Syndrome, 3mc Syndrome	AR	100	11 of 11
<b>COMT</b>	22q11.2 Deletion Syndrome	AD	99.98	5 of 5
<b>CTCF</b>	Intellectual Disability-Feeding Difficulties-Developmental Delay-Microcephaly Syndrome	AD	96.6	39 of 41
<b>CWC27</b>	Retinitis Pigmentosa With Or Without Skeletal Anomalies	AR	99.77	8 of 8
<b>CYP26B1</b>	Craniosynostosis With Radiohumeral Fusions And Other Skeletal And Craniofacial Anomalies	AR	100	12 of 12
<b>DMP1</b>	Hypophosphatemic Rickets	AR	99.89	11 of 11
<b>DONSON</b>	Microcephaly-Micromelia Syndrome, Microcephaly, Short Stature, And Limb Abnormalities	AR	98.14	26 of 27
<b>DPF2</b>	Coffin-Siris Syndrome	AD	99.99	10 of 10



<b>DPH1</b>	Developmental Delay With Short Stature, Dysmorphic Features, And Sparse Hair, Craniofacial Dysplasia-Short Stature-Ectodermal Anomalies-Intellectual Disability Syndrome	AR	100	8 of 8
<b>DSE</b>	Musculocontractural Ehlers-Danlos Syndrome	AR	99.94	3 of 3
<b>EDNRB</b>	Abcd Syndrome, Waardenburg-Shah Syndrome	AD,AR	99.55	70 of 72
<b>EFNB1</b>	Craniofrontonasal Syndrome	X,XD,G	100	-
<b>ENPP1</b>	Cole Disease, Hypophosphatemic Rickets	AD,AR,MU,P	96.59	73 of 75
<b>ERCC2</b>	Cerebrooculofacioskeletal Syndrome, Trichothiodystrophy, Xeroderma Pigmentosum-Cockayne Syndrome Complex	AR	100	102 of 102
<b>ERCC3</b>	Trichothiodystrophy, Xeroderma Pigmentosum-Cockayne Syndrome Complex	AR	99.98	24 of 24
<b>ERF</b>	Chitayat Syndrome, Lambdoid Synostosis, Crouzon Disease, Isolated Cloverleaf Skull Syndrome, Isolated Scaphocephaly	AD	99.73	31 of 31
<b>ESCO2</b>	Roberts Syndrome, Sc Phocomelia Syndrome	AR	99.69	32 of 32
<b>ETS2</b>	Down Syndrome		99.99	1 of 2
<b>EXTL3</b>	Immunoskeletal Dysplasia With Neurodevelopmental Abnormalities Skeletal Dysplasia-T-cell Immunodeficiency-Developmental Delay Syndrome	AR	99.99	10 of 10
<b>FAT4</b>	Hennekam Lymphangiectasia-lymphedema Syndrome, Van Maldergem Syndrome, Cerebrofacioarticular Syndrome	AR	99.8	41 of 41
<b>FBN1</b>	Acromicric Dysplasia, Isolated Ectopia Lentis, Geleophysic Dysplasia, Marfan Lipodystrophy Syndrome, Mass Syndrome, Weill-Marchesani Syndrome, Acromicric Dysplasia, Shprintzen-Goldberg Syndrome	AD	100	2836 of 2845
<b>FGF9</b>	Multiple Synostoses Syndrome	AD	100	2 of 2
<b>FGFR1</b>	Encephalocraniocutaneous Lipomatosis, Hartsfield Syndrome, Jackson-Weiss Syndrome, Osteoglyphonic Dysplasia, Pfeiffer Syndrome, Nonsyndromic Trigenocephaly, Encephalocraniocutaneous Lipomatosis, Lobar Holoprosencephaly, Microform Holoprosencephaly, Septo-Optic Dysplasia Spectrum	AD	100	279 of 280
<b>FGFR2</b>	Antley-Bixler Syndrome Without Genital Anomalies Or Disordered Steroidogenesis, Apert Syndrome, Bent Bone Dysplasia Syndrome, Crouzon Syndrome, Cutis Gyrata Syndrome Of Beare And Stevenson, Familial Scaphocephaly Syndrome,, Lacrimoauriculodentodigital Syndrome, Pfeiffer Syndrome, Saethre-Chotzen Syndrome, Familial Scaphocephaly Syndrome, Fgfr2-Related Bent Bone Dysplasia	AD	98	140 of 143
<b>FGFR3</b>	Achondroplasia With Developmental Delay And Acanthosis Nigricans, Camptodactyly, Tall Stature, And Hearing Loss Syndrome, Hypochondroplasia, Lacrimoauriculodentodigital Syndrome, Muenke Syndrome, Thanatophoric Dysplasia, Isolated Brachycephaly, Isolated Plagiocephaly, Saethre-Chotzen Syndrome	AD,AR	99.89	77 of 78
<b>FLNA</b>	Fg Syndrome, Frontometaphyseal Dysplasia, Melnick-Needles Syndrome, Otopalatodigital Syndrome, Terminal Osseous Dysplasia, Frontometaphyseal Dysplasia, Otopalatodigital Syndrome, X-linked Ehlers-Danlos Syndrome	X,XR,XD,G	100	-
<b>FLNB</b>	Atelosteogenesis, Boomerang Dysplasia, Larsen Syndrome, Spondyllocarpotarsal Synostosis Syndrome	AD,AR	100	124 of 124
<b>FREM1</b>	Manitoba Oculotrichoanal Syndrome, Trigenocephaly, Bnar Syndrome, Isolated Trigenocephaly	AD,AR	97.32	27 of 30
<b>GDF5</b>	Acromesomelic Dysplasia, Brachydactyly, Chondrodysplasia, Fibular Hypoplasia And Complex Brachydactyly, Multiple Synostoses Syndrome, Symphalangism, Angel-Shaped Phalango-Epiphyseal Dysplasia, Brachydactyly Syndrome, Multiple Synostoses Syndrome, Proximal Symphalangism	AD,AR	99.48	48 of 51
<b>GLI3</b>	Greig Cephalopolysyndactyly Syndrome, Pallister-Hall Syndrome, Polydactyly, Acrocallosal Syndrome,Tibial Hemimelia	AD,AR	100	231 of 231
<b>GLIS3</b>	Diabetes Mellitus, Neonatal, With Congenital Hypothyroidism	AR	99.83	21 of 21
<b>GMNN</b>	Meier-Gorlin Syndrome, Ear-Patella-Short Stature Syndrome	AD	99.72	3 of 3
<b>GNPTAB</b>	Mucopolipidosis II and III	AR	100	279 of 280
<b>GP1BB</b>	22q11.2 Deletion Syndrome	AR	74.08	26 of 50
<b>GPC6</b>	Autosomal Recessive Omodysplasia	AR	99.92	3 of 3
<b>GTF2E2</b>	Nonphotosensitive Trichothiodystrophy	AR	99.98	2 of 2
<b>GTF2H5</b>	Photosensitive Trichothiodystrophy	AR	100	8 of 8
<b>HIRA</b>	22q11.2 Deletion Syndrome	-	99.99	5 of 5
<b>HNRNPK</b>	Au-Kline Syndrome; Auks ,	AD	99.88	16 of 17
<b>HUWE1</b>	X-linked Intellectual Disability	X,G	99.41	-
<b>IFT122</b>	Cranioectodermal Dysplasia	AR	99.83	22 of 22



<b>IFT140</b>	Short-Rib Thoracic Dysplasia With Or Without Polydactyly, Jeune Syndrome	AR	99.97	81 of 81
<b>IFT43</b>	Cranioectodermal Dysplasia, Shoty-Rib Thoracic Dysplasia With Polydactyly	AR	100	6 of 6
<b>IFT52</b>	Short-Rib Thoracic Dysplasia With Or Without Polydactyly, Cranioectodermal Dysplasia	AR	99.8	8 of 8
<b>IHH</b>	Acrocapitofemoral Dysplasia, Brachydactyly, Acrocapitofemoral Dysplasia	AD,AR	99.39	28 of 29
<b>IL11RA</b>	Craniosynostosis And Dental Anomalies	AR	100	22 of 22
<b>IL6ST</b>	Hyper-IgE Recurrent Infection Syndrome	AR	99.34	2 of 2
<b>IRX5</b>	Hamamy Syndrome	AR	97.1	5 of 5
<b>JMJD1C</b>	22q11.2 Deletion Syndrome		99.09	27 of 27
<b>KAT6A</b>	Arboleda-Tham Syndrome, Autosomal Dominant Intellectual Disability-Craniofacial Anomalies-Cardiac Defects Syndrome	AD	99.89	66 of 68
<b>KPTN</b>	Macrocephaly-Developmental Delay Syndrome	AR	100	5 of 5
<b>KRAS</b>	Aplasia Cutis Congenita With Epibulbar Dermoids, Cardiofaciocutaneous Syndrome, Noonan Syndrome, Schimmelpenning-Feuerstein-Mims Syndrome, Encephalocraniocutaneous Lipomatosis, Toriello-Lacassie-Droste Syndrome	AD	100	38 of 38
<b>LEMD3</b>	Buschke-Ollendorff Syndrome, 12q14 Microdeletion Syndrome, Isolated Osteopoikilosis, Melorheostosis With Osteopoikilosis	AD	99.06	30 of 33
<b>LIG4</b>	Lig4 Syndrome, Dubowitz Syndrome, Omenn Syndrome	AR	99.48	46 of 46
<b>LRP5</b>	Endosteal Hyperostosis, Osteopetrosis, Van Buchem Disease, Endosteal Hyperostosis, Hyperostosis Corticalis Generalisata, Osteosclerosis-Developmental Delay-Craniosynostosis Syndrome	AD,AR	98.12	265 of 269
<b>MAF</b>	Ayme-gripp Syndrome, Cataract-Microcornea Syndrome	AD	75.14	23 of 23
<b>MAN2B1</b>	Alpha-Mannosidosis	AR	100	149 of 149
<b>MAP3K7</b>	Cardiospondylocarpofacial Syndrome, Frontometaphyseal Dysplasia	AD	99.96	13 of 13
<b>MASP1</b>	3mc Syndrome	AR	100	29 of 30
<b>MED12</b>	Lujan-Fryns Syndrome, Ohdo Syndrome, Opitz-Kaveggia Syndrome, Fg Syndrome, X-linked Intellectual Disability With Marfanoid Habitus	X,XR,G	100	-
<b>MEGF8</b>	Carpenter Syndrome	AR	98.97	22 of 22
<b>MIR140</b>	Spondyloepiphyseal Dysplasia	AD	-	-
<b>MPLKIP</b>	Nonphotosensitive Trichothiodystrophy	AR	100	13 of 13
<b>MSX2</b>	Craniosynostosis, Parietal Foramina With Cleidocranial Dysplasia, Enlarged Parietal Foramina	AD	99.98	15 of 15
<b>MYH3</b>	Arthrogyrosis, Contractures, Pterygia, And Spondylocarpostarsal Fusion Syndrome, Autosomal Recessive Multiple Pterygium Syndrome, Digitotalar Dysmorphism, Freeman-Sheldon Syndrome, Sheldon-Hall Syndrome	AD,AR	100	46 of 47
<b>NFIX</b>	Marshall-Smith Syndrome, Sotos Syndrome, 19p13.3 Microduplication Syndrome, Malan Overgrowth Syndrome, Marshall-Smith Syndrome	AD	94.42	75 of 81
<b>NOG</b>	Brachydactyly, Multiple Synostoses Syndrome, Stapes Ankylosis With Broad Thumb And Toes, Symphalangism, Tarsal-Carpal Coalition Syndrome, Synostosis Of Talus And Calcaneus With Short Stature	AD	99.89	61 of 62
<b>NSD1</b>	Sotos Syndrome, 5q35 Microduplication Syndrome, Weaver Syndrome	AD	99.8	451 of 459
<b>NSUN2</b>	Autosomal Recessive Non-Syndromic Intellectual Disability, Dubowitz Syndrome	AR	99.99	8 of 8
<b>ORC1</b>	Ear, Patella, Short Stature Syndrome	AR	100	12 of 12
<b>ORC4</b>	Meier-Gorlin Syndrome, Ear-Patella-Short Stature Syndrome	AR	100	4 of 4
<b>ORC6</b>	Meier-Gorlin Syndrome, Ear-Patella-Short Stature Syndrome	AR	100	6 of 6
<b>P4HB</b>	Cole-Carpenter Syndrome	AD	94.97	13 of 13
<b>PAX3</b>	Craniofacial-Deafness-Hand Syndrome, Waardenburg Syndrome	AD,AR	99.98	157 of 157
<b>PCNT</b>	Microcephalic Osteodysplastic Primordial Dwarfism, Seckel Syndrome	AR	99.92	103 of 105
<b>PHEX</b>	X-linked Hypophosphatemia	X,XD,G	99.42	-
<b>PIGT</b>	Multiple Congenital Anomalies-Hypotonia-Seizures Syndrome, Paroxysmal Nocturnal Hemoglobinuria, Intellectual Disability-Seizures-Hypophosphatasia-Ophthalmic-Skeletal Anomalies Syndrome	AD,AR	100	15 of 15
<b>PLK4</b>	Microcephaly And Chorioretinopathy, Seckel Syndrome	AR	99.74	10 of 10
<b>POLA1</b>	Van Esch-O'driscoll Syndrome, X-linked Intellectual Disability	X,XR,G	99.26	-
<b>POR</b>	Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis	AD,AR	99.98	67 of 68
<b>PPP1CB</b>	Noonan Syndrome-Like Disorder With Loose Anagen Hair	AD	99.87	12 of 12
<b>PPP3CA</b>	Arthrogyrosis, Cleft Palate, Craniosynostosis, And Impaired Intellectual Development, Undetermined Early-Onset Epileptic Encephalopathy	AD	99.98	16 of 16



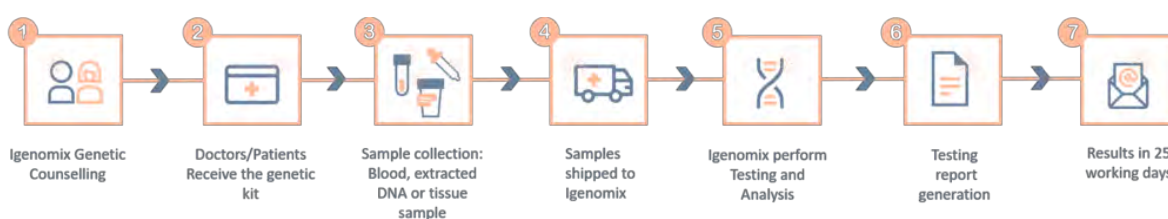
<b>PSAT1</b>	Neu-Laxova Syndrome, Phosphoserine Aminotransferase Deficiency	AR	99.95	9 of 9
<b>PTEN</b>	Macrocephaly/Autism Syndrome, Bannayan-Riley-Ruvalcaba Syndrome, Lhermitte-Duclos Disease, Segmental Outgrowth-Lipomatosis-Arteriovenous Malformation-Epidermal Nevus Syndrome	AD	99.97	609 of 629
<b>RAB23</b>	Carpenter Syndrome	AR	100	15 of 15
<b>RAC3</b>	Neurodevelopmental Disorder With Structural Brain Anomalies And Dysmorphic Facies	AD	94.13	5 of 5
<b>RBBP8</b>	Jawad Syndrome, Seckel Syndrome	AR	96.02	6 of 6
<b>RECQL4</b>	Baller-Gerold Syndrome, Rapadilino Syndrome, Rothmund-Thomson Syndrome	AR	96.72	134 of 135
<b>RNF113A</b>	Nonphotosensitive Trichothiodystrophy	X,XD,G	99.7	-
<b>RREB1</b>	22q11.2 Deletion Syndrome		99.92	8 of 8
<b>RSPRY1</b>	Spondyloepimetaphyseal Dysplasia	AR	99.98	4 of 4
<b>RTTN</b>	Microcephaly, Short Stature, And Polymicrogyria With Seizures, Microcephalic Primordial Dwarfism Due To Rttm Deficiency	AR	99.94	28 of 29
<b>RUNX2</b>	Cleidocranial Dysplasia, Metaphyseal Dysplasia With Maxillary Hypoplasia And Brachydactyly, Cleidocranial Dysplasia	AD	73.67	189 of 190
<b>SCARF2</b>	Van Den Ende-Gupta Syndrome	AR	93.06	13 of 13
<b>SEC24C</b>	22q11.2 Deletion Syndrome		99.98	-
<b>SEC24D</b>	Cole-Carpenter Syndrome	AR	99.97	14 of 14
<b>SETD2</b>	Luscan-Lumish Syndrome, Sotos Syndrome	AD	99.83	19 of 19
<b>SKI</b>	Shprintzen-Goldberg Craniosynostosis Syndrome, 1p36 Deletion Syndrome	AD	99.66	39 of 39
<b>SLC12A6</b>	Corpus Callosum Agenesis-Neuronopathy Syndrome	AR	100	21 of 21
<b>SLC25A24</b>	Fontaine Progeroid Syndrome, Gorlin-Chaudhry-Moss Syndrome	AD	99.59	2 of 2
<b>SLC2A10</b>	Arterial Tortuosity Syndrome	AR	100	35 of 35
<b>SLC35A2</b>	Congenital Disorder Of Glycosylation	X,XD,G	99.97	-
<b>SLC39A8</b>	Congenital Disorder Of Glycosylation	AR	99.89	7 of 7
<b>SMAD3</b>	Loeys-Dietz Syndrome, Aneurysm-Osteoarthritis Syndrome	AD	100	128 of 128
<b>SMAD6</b>	Craniosynostosis	AD	80.88	64 of 74
<b>SMO</b>	Craniofacial Malformations With Polysyndactyly And Abnormal Skin And Gut Development, Curry-Jones Syndrome	AR	94.03	10 of 10
<b>SNX10</b>	Autosomal Recessive Malignant Osteopetrosis	AR	100	14 of 14
<b>SON</b>	Zttk Syndrome, Brain Malformations-Musculoskeletal Abnormalities-Facial Dysmorphism-Intellectual Disability Syndrome	AD	99.27	30 of 32
<b>SOX10</b>	Peripheral Demyelinating Neuropathy, Central Dysmyelination, Waardenburg Syndrome	AD	99.74	139 of 147
<b>SPECC1L</b>	Facial Clefting, Hypertelorism, Opitz Gbbb Syndrome	AD	99.66	14 of 14
<b>STAT3</b>	Hyper-IgE Recurrent Infection Syndrome, Permanent Neonatal Diabetes Mellitus	AD	100	171 of 171
<b>TANC2</b>	Intellectual Developmental Disorder With Autistic Features And Language Delay, With Or Without Seizures, Non-Specific Syndromic Intellectual Disability	AD	97.81	21 of 21
<b>TARS1</b>	Nonphotosensitive Trichothiodystrophy	AR	99.94	-
<b>TBC1D24</b>	Doors Syndrome, Rolandic Epilepsy With Paroxysmal Exercise-Induced Dystonia Andwriter's Cramp, Myoclonic Epilepsy	AD,AR	100	80 of 80
<b>TBX1</b>	DiGeorge Syndrome, Velocardiofacial Syndrome	AD,AR	88.7	35 of 42
<b>TCF12</b>	Craniosynostosis, Isolated Brachycephaly, Isolated Plagiocephaly	AD	99.98	73 of 76
<b>TCIRG1</b>	Osteopetrosis, Dysosteosclerosis	AR	100	140 of 146
<b>TCOF1</b>	Treacher Collins-Franceschetti Syndrome	AD	100	326 of 327
<b>TGFB2</b>	Loeys-Dietz Syndrome	AD	99.9	41 of 44
<b>TGFB3</b>	Loeys-Dietz Syndrome	AD	100	34 of 35
<b>TGFBR1</b>	Loeys-Dietz Syndrome	AD	94	96 of 100
<b>TGFBR2</b>	Loeys-Dietz Syndrome	AD	99.9	165 of 166
<b>TLK2</b>	Autosomal Dominant Mental Retardation	AD	96.98	39 of 39
<b>TMCO1</b>	Cerebrofaciothoracic Dysplasia	AR	88	5 of 5
<b>TNFSF11</b>	Autosomal Recessive Malignant Osteopetrosis	AR	99.84	4 of 4
<b>TRAIP</b>	Seckel Syndrome	AR	100	2 of 2
<b>TWIST1</b>	Craniosynostosis, Robinow-Sorauf Syndrome, Saethre-Chotzen Syndrome, Sweeney-Cox Syndrome, Isolated Brachycephaly, Isolated Plagiocephaly, Isolated Scaphocephaly	AD	74.06	133 of 161
<b>TWIST2</b>	Ablepharon-Macrostomia Syndrome, Barber-Say Syndrome	AD,AR	99.82	9 of 9
<b>UFD1</b>	22q11.2 Deletion Syndrome	-	99.98	-
<b>WDR19</b>	Cranioectodermal Dysplasia, Senior-Loken Syndrome, Jeune Syndrome	AR	99.96	47 of 49

<b>WDR35</b>	Cranioectodermal Dysplasia, Short Rib-Polydactyly Syndrome	AR	100	31 of 33
<b>YY1</b>	Gabriele-de Vries Syndrome	AD	99.89	13 of 13
<b>ZEB2</b>	Mowat-Wilson Syndrome	AD	98.95	253 of 254
<b>ZIC1</b>	Craniosynostosis, Structural Brain Anomalies With Impaired Intellectual Development And Craniosynostosis, Isolated Brachycephaly, Isolated Oxycephaly, Isolated Plagiocephaly	AD	100	7 of 7

\*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](mailto: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. Delahaye S, Bernard JP, Rénier D, Ville Y. Prenatal ultrasound diagnosis of fetal craniosynostosis. *Ultrasound Obstet Gynecol.* 2003 Apr;21(4):347-53. doi: 10.1002/uog.91. PMID: 12704742
2. Kajdic, N., Spazzapan, P., & Velnar, T. (2018). Craniosynostosis - Recognition, clinical characteristics, and treatment. *Bosnian journal of basic medical sciences*, 18(2), 110–116. <https://doi.org/10.17305/bjbm.2017.2083>
3. Wilkie, A., Johnson, D., & Wall, S. A. (2017). Clinical genetics of craniosynostosis. *Current opinion in pediatrics*, 29(6), 622–628. <https://doi.org/10.1097/MOP.0000000000000542>
4. Kutkowska-Kaźmierczak, A., Gos, M., & Obersztyn, E. (2018). Craniosynostosis as a clinical and diagnostic problem: molecular pathology and genetic counseling. *Journal of applied genetics*, 59(2), 133–147. <https://doi.org/10.1007/s13353-017-0423-4>
5. Wang, J. C., Nagy, L., & Demke, J. C. (2016). Syndromic Craniosynostosis. *Facial plastic surgery clinics of North America*, 24(4), 531–543. <https://doi.org/10.1016/j.fsc.2016.06.008>
6. Azoulay-Avinoam, S., Bruun, R., MacLaine, J., Allareddy, V., Resnick, C. M., & Padwa, B. L. (2020). An Overview of Craniosynostosis Craniofacial Syndromes for Combined Orthodontic and Surgical Management. *Oral and maxillofacial surgery clinics of North America*, 32(2), 233–247. <https://doi.org/10.1016/j.coms.2020.01.004>
7. Jabs, E. (2008). Toward understanding the pathogenesis of craniosynostosis through clinical and molecular correlates. *Clinical Genetics*, 53(2), 79-86. doi: 10.1111/j.1399-0004.1998.tb02648.x
8. Kimonis, V., Gold, J., Hoffman, T., Panchal, J., & Boyadjiev, S. (2007). Genetics of Craniosynostosis. *Seminars In Pediatric Neurology*, 14(3), 150-161. doi: 10.1016/j.spen.2007.08.008
9. Lattanzi, W., Barba, M., Di Pietro, L., & Boyadjiev, S. A. (2017). Genetic advances in craniosynostosis. *American journal of medical genetics. Part A*, 173(5), 1406–1429. <https://doi.org/10.1002/ajmg.a.38159>