



# 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

ADAMTS    Hennekam Syndrome	GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
AKT1 Proteus Syndrome, Meningioma AD 100 6 of 6	ADAMTS3	Hennekam Syndrome	AR	99.97	4 of 4
ALPL         Hypophosphatasia         AD,AR         100         320 of 321           ALX3         Frontonasal Dysplasia         AR         89.31         8 of 8           ALX4         Craniosynostosis, Frontonasal Dysplasia, Enlarged Parietal Foramina, 100         AD,AR         89.94         25 of 25           APC2         Complex Cortical Dysplasia, Sotos Syndrome         AR         99.95         2 of 25           ASX11         Bohring-Opitz Syndrome, Bohring-Opitz Syndrome         AD         99.95         2 of 2           ASX11         Bohring-Opitz Syndrome, Bohring-Opitz Syndrome         AD AD         99.98         39 of 40           ATR         Cattaneous Telanglectasia And Cancer Syndrome, Seckel Syndrome         AD AD         89.98         39 of 40           ATRIP         Seckel Syndrome         AD         AP.99.89         2 of 2           BBGAT3         With Or Without Congenital Heart Defects         AR         99.86         15 of 15           BBGCT         Peters Plus Syndrome         AP.         99.86         15 of 15           BBGACT         Peters Plus Syndrome         AP.         99.89         10 of 15           CCBE1         Helier-Borno Syndrome, Syndrome, Spondylodysplastic Type         AR         99.96         10 of 15           BWAGATA	AHDC1		AD	99.87	41 of 43
ALXA Frontonasal Dysplasia AR 89.31 8 of 8  ALXA Frontonasal Dysplasia Enlarged Parletal Foramina, Isolated Scaphocephaly, Potock-Shaffer Syndrome  APC2 Complex Cortical Dysplasia, Stotos Syndrome  APC2 Complex Cortical Dysplasia, Stotos Syndrome  AR 94.97 11 of 11  ARVCF 22q1.1.2 Deletion Syndrome, Bohring-Opitz Syndrome  ARXIL Bohring-Opitz Syndrome, Spondylodysplasia Type  BAGAT3 With Or Without Congenital Heart Defects  BAGAT3 With Or Without Congenital Heart Defects  BAGAT4 With Or Without Congenital Heart Defects  BAGAT5 With Or Without Congenital Heart Defects  BAPT2 Chondrodysplasia With Joint Dislocations  AR 99.96 -  Ehlers-Danlos Syndrome, Spondylodysplastic Type  BAPT2 Chondrodysplasia With Joint Dislocations  AR 99.96 -  Ehlers-Danlos Syndrome, Spondylodysplastic Type  BAPT2 Chondrodysplasia With Joint Dislocations  AR 99.97 100 38 of 42  CCBE1 Hennekan Lymphangiectasia-Lymphedema Syndrome  CRB1 Hennekan Lymphangiectasia-Lymphedema Syndrome  CRB1 Hennekan Lymphangiectasia-Lymphedema Syndrome  CRB1 Meier-Gorlin Syndrome, Ear-Patella-Short Stature Syndrome  AR 99.99 19 of 19  CDG6 Meier-Gorlin Syndrome, Ear-Patella-Short Stature Syndrome  AR 99.99 19 of 19  CDG6 Meier-Gorlin Syndrome, Ear-Patella-Short Stature Syndrome  AR 99.99 10 of 10  Meier-Gorlin Syndrome, Ear-Patella-Short Stature Syndrome  AR 99.99 10 of 10  Meier-Gorlin Syndrome, Ear-Patella-Short Stature Syndrome  AR 99.99 10 of 10  Meier-Gorlin Syndrome, Ear-Patella-Short Stature Syndrome  AR 99.99 10 of 10  Meier-Gorlin Syndrome, Ear-Patella-Short Stature Syndrome  AR 99.99 10 of 10  Meier-Gorlin Syndrome, Ear-Patella-Short Stature Syndrome  AR 99.91 10 of 10  CDT1 Meier-Gorlin Syndrome, Ear-Patella-Short Stature Syndrome  AR 99.99 10 of 10  Meier-Gorlin Syndrome, Ear-Patell	AKT1	Proteus Syndrome, Meningioma	AD	100	
ALX4   Craniosynostosis, Frontonasal Dysplasia, Enlarged Parietal Foramina, Isolated Scaphocephaly, Potocki-Shaffer Syndrome	ALPL	Hypophosphatasia	AD,AR	100	
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CYP26B1Craniosynostosis With Radiohumeral Fusions And Other Skeletal And Craniofacial AnomaliesAR10012 of 12DMP1Hypophosphatemic RicketsAR99.8911 of 11DONSONMicrocephaly-Micromelia Syndrome, Microcephaly, Short Stature, And Limb AbnormalitiesAR98.1426 of 27	CTCF		AD	96.6	39 of 41
CYP26B1 Craniofacial Anomalies AR 100 12 of 12  DMP1 Hypophosphatemic Rickets AR 99.89 11 of 11  DONSON Microcephaly-Micromelia Syndrome, Microcephaly, Short Stature, And Limb Abnormalities AR 98.14 26 of 27	CWC27	Retinitis Pigmentosa With Or Without Skeletal Anomalies	AR	99.77	8 of 8
DMP1Hypophosphatemic RicketsAR99.8911 of 11DONSONMicrocephaly-Micromelia Syndrome, Microcephaly, Short Stature, And Limb AbnormalitiesAR98.1426 of 27	CYP26B1	· ·	AR	100	12 of 12
DONSONMicrocephaly-Micromelia Syndrome, Microcephaly, Short Stature, And Limb AbnormalitiesAR98.1426 of 27	DMP1		AR	99.89	11 of 11
		Microcephaly-Micromelia Syndrome, Microcephaly, Short Stature, And			
	DPF2		AD	99.99	10 of 10





	Developmental Delay With Short Stature, Dysmorphic Features, And			
DPH1	Sparse Hair, Craniofacial Dysplasia-Short Stature-Ectodermal Anomalies-	AR	100	8 of 8
DCF	Intellectual Disability Syndrome	A D	00.04	2 of 2
DSE EDNRB	Musculocontractural Ehlers-Danlos Syndrome Abcd Syndrome, Waardenburg-Shah Syndrome	AR	99.94 99.55	3 of 3 70 of 72
EFNB1		AD,AR X,XD,G	100	70 01 72
ENPP1	Craniofrontonasal Syndrome Cole Disease, Hypophosphatemic Rickets	AD,AR,MU,P	96.59	- 73 of 75
ENPPI	Cerebrooculofacioskeletal Syndrome, Trichothiodystrophy, Xeroderma	AD,AR,IVIO,P	90.59	
ERCC2	Pigmentosum-Cockayne Syndrome Complex	AR	100	102 of 102
ERCC3	Trichothiodystrophy, Xeroderma Pigmentosum-Cockayne Syndrome Complex	AR	99.98	24 of 24
ERF	Chitayat Syndrome, Lambdoid Synostosis, Crouzon Disease, Isolated Cloverleaf Skull Syndrome, Isolated Scaphocephaly	AD	99.73	31 of 31
ESCO2	Roberts Syndrome, Sc Phocomelia Syndrome	AR	99.69	32 of 32
ETS2	Down Syndrome		99.99	1 of 2
	Immunoskeletal Dysplasia With Neurodevelopmental Abnormalities			
EXTL3	Skeletal Dysplasia-T-cell Immunodeficiency-Developmental Delay Syndrome	AR	99.99	10 of 10
FAT4	Hennekam Lymphangiectasia-lymphedema Syndrome, Van Maldergem Syndrome, Cerebrofacioarticular Syndrome	AR	99.8	41 of 41
FBN1	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
FGF9	Multiple Synostoses Syndrome	AD	100	2 of 2
10.5	Encephalocraniocutaneous Lipomatosis, Hartsfield Syndrome, Jackson-	AD	100	2012
FGFR1	Weiss Syndrome, Osteoglophonic Dysplasia, Pfeiffer Syndrome, Nonsyndromic Trigonocephaly, Encephalocraniocutaneous Lipomatosis, Lobar Holoprosencephaly, Microform Holoprosencephaly, Septo-Optic Dysplasia Spectrum	AD	100	279 of 280
FGFR2	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
FGFR3	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
FLNA	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	-
FLNB	Atelosteogenesis, Boomerang Dysplasia, Larsen Syndrome, Spondylocarpotarsal Synostosis Syndrome	AD,AR	100	124 of 124
FREM1	Manitoba Oculotrichoanal Syndrome, Trigonocephaly, Bnar Syndrome,	AD,AR	97.32	27 of 30
GDF5	Isolated Trigonocephaly 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
GLI3	Greig Cephalopolysyndactyly Syndrome, Pallister-Hall Syndrome, Polydactyly, Acrocallosal Syndrome,Tibial Hemimelia	AD,AR	100	231 of 231
GLIS3	Diabetes Mellitus, Neonatal, With Congenital Hypothyroidism	AR	99.83	21 of 21
GMNN	Meier-Gorlin Syndrome, Ear-Patella-Short Stature Syndrome	AD	99.72	3 of 3
GNPTAB	Mucolipidosis II and III	AR	100	279 of
GP1BB	22q11.2 Deletion Syndrome	AR	74.08	280 26 of 50
GPC6	Autosomal Recessive Omodysplasia	AR	99.92	3 of 3
GTF2E2	Nonphotosensitive Trichothiodystrophy	AR	99.98	2 of 2
GTF2H5	Photosensitive Trichothiodystrophy	AR	100	8 of 8
HIRA	22q11.2 Deletion Syndrome	-	99.99	5 of 5
HNRNPK	Au-Kline Syndrome; Auks ,	AD	99.88	16 of 17
HUWE1	X-linked Intellectual Disability	X,G	99.88	10 01 17
	·	·		22 of 22
IFT122	Cranioectodermal Dysplasia	AR	99.83	22 of 22





IFT140	Short-Rib Thoracic Dysplasia With Or Without Polydactyly, Jeune Syndrome	AR	99.97	81 of 81
IFT43	Cranioectodermal Dysplasia, Shoty-Rib Thoracic Dysplasia With Polydactyly	AR	100	6 of 6
IFT52	Short-Rib Thoracic Dysplasia With Or Without Polydactyly, Cranioectodermal Dysplasia	AR	99.8	8 of 8
IHH	Acrocapitofemoral Dysplasia, Brachydactyly, Acrocapitofemoral Dysplasia	AD,AR	99.39	28 of 29
IL11RA	Craniosynostosis And Dental Anomalies	AR	100	22 of 22
IL6ST IRX5	Hyper-IgE Recurrent Infection Syndrome Hamamy Syndrome	AR AR	99.34 97.1	2 of 2 5 of 5
JMJD1C	22q11.2 Deletion Syndrome	AN	99.09	27 of 27
KAT6A	Arboleda-Tham Syndrome, Autosomal Dominant Intellectual Disability-	AD	99.89	66 of 68
KPTN	Craniofacial Anomalies-Cardiac Defects Syndrome  Macrocephaly-Developmental Delay Syndrome	AR	100	5 of 5
KRAS	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
LEMD3	Buschke-Ollendorff Syndrome, 12q14 Microdeletion Syndrome, Isolated Osteopoikilosis, Melorheostosis With Osteopoikilosis	AD	99.06	30 of 33
LIG4	Lig4 Syndrome, Dubowitz Syndrome, Omenn Syndrome	AR	99.48	46 of 46
LRP5	Endosteal Hyperostosis, Osteopetrosis, Van Buchem Disease, Endosteal Hyperostosis, Hyperostosis Corticalis Generalisata, Osteosclerosis-Developmental Delay-Craniosynostosis Syndrome	AD,AR	98.12	265 of 269
MAF	Ayme-gripp Syndrome, Cataract-Microcornea Syndrome	AD	75.14	23 of 23
MAN2B1	Alpha-Mannosidosis	AR	100	149 of 149
МАРЗК7	Cardiospondylocarpofacial Syndrome, Frontometaphyseal Dysplasia	AD	99.96	13 of 13
MASP1	3mc Syndrome	AR	100	29 of 30
MED12	Lujan-Fryns Syndrome, Ohdo Syndrome, Opitz-Kaveggia Syndrome, Fg Syndrome, X-linked Intellectual Disability With Marfanoid Habitus	X,XR,G	100	-
MEGF8	Carpenter Syndrome	AR	98.97	22 of 22
MIR140 MPLKIP	Spondyloepiphyseal Dysplasia Nonphotosensitive Trichothiodystrophy	AD AR	100	- 13 of 13
	Craniosynostosis, Parietal Foramina With Cleidocranial Dysplasia,			
MSX2	Enlarged Parietal Foramina	AD	99.98	15 of 15
МҮН3	Arthrogryposis, 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
NFIX	Marshall-Smith Syndrome, Sotos Syndrome, 19p13.3 Microduplication Syndrome, Malan Overgrowth Syndrome, Marshall-Smith Syndrome	AD	94.42	75 of 81
NOG	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
NSD1	Sotos Syndrome, 5q35 Microduplication Syndrome, Weaver Syndrome	AD	99.8	451 of 459
NSUN2	Autosomal Recessive Non-Syndromic Intellectual Disability, Dubowitz Syndrome	AR	99.99	8 of 8
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 P4HB	Meier-Gorlin Syndrome, Ear-Patella-Short Stature Syndrome Cole-Carpenter Syndrome	AR AD	100 94.97	6 of 6 13 of 13
PAX3	Craniofacial-Deafness-Hand Syndrome, Waardenburg Syndrome	AD,AR	99.98	157 of 157
PCNT	Microcephalic Osteodysplastic Primordial Dwarfism, Seckel Syndrome	AR	99.92	103 of 105
PHEX	X-linked Hypophosphatemia	X,XD,G	99.42	-
PIGT	Multiple Congenital Anomalies-Hypotonia-Seizures Syndrome, Paroxysmal Nocturnal Hemoglobinuria, Intellectual Disability-Seizures- Hypophosphatasia-Ophthalmic-Skeletal Anomalies Syndrome	AD,AR	100	15 of 15
PLK4	Microcephaly And Chorioretinopathy, Seckel Syndrome	AR	99.74	10 of 10
POLA1	Van Esch-O'driscoll Syndrome, X-linked Intellectual Disability	X,XR,G	99.26	-
POR	Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis	AD,AR	99.98	67 of 68
PPP1CB	Noonan Syndrome-Like Disorder With Loose Anagen Hair	AD	99.87	12 of 12
PPP3CA	Arthrogryposis, Cleft Palate, Craniosynostosis, And Impaired Intellectual Development, Undetermined Early-Onset Epileptic Encephalopathy	AD	99.98	16 of 16





PSAT1	Neu-Laxova Syndrome, Phosphoserine Aminotransferase Deficiency	AR	99.95	9 of 9
	Macrocephaly/Autism Syndrome, Bannayan-Riley-Ruvalcaba Syndrome,			609 of
PTEN	Lhermitte-Duclos Disease, Segmental Outgrowth-Lipomatosis-	AD	99.97	629
D 4 D 2 2	Arteriovenous Malformation-Epidermal Nevus Syndrome	A.D.	100	45 - 545
RAB23	Carpenter Syndrome Neurodevelopmental Disorder With Structural Brain Anomalies And	AR	100	15 of 15
RAC3	Dysmorphic Facies	AD	94.13	5 of 5
RBBP8	Jawad Syndrome, Seckel Syndrome	AR	96.02	6 of 6
	Baller-Gerold Syndrome, Rapadilino Syndrome, Rothmund-Thomson			134 of
RECQL4	Syndrome	AR	96.72	135
RNF113A	Nonphotosensitive Trichothiodystrophy	X,XD,G	99.7	-
RREB1	22q11.2 Deletion Syndrome		99.92	8 of 8
RSPRY1	Spondyloepimetaphyseal Dysplasia	AR	99.98	4 of 4
RTTN	Microcephaly, Short Stature, And Polymicrogyria With Seizures,	AR	99.94	28 of 29
	Microcephalic Primordial Dwarfism Due To Rttn Deficiency			100 of
RUNX2	Cleidocranial Dysplasia, Metaphyseal Dysplasia With Maxillary Hypoplasia And Brachydactyly, Cleidocranial Dysplasia	AD	73.67	189 of 190
SCARF2	Van Den Ende-Gupta Syndrome	AR	93.06	13 of 13
SEC24C	22q11.2 Deletion Syndrome	All	99.98	-
SEC24D	Cole-Carpenter Syndrome	AR	99.97	14 of 14
SETD2	Luscan-Lumish Syndrome, Sotos Syndrome	AD	99.83	19 of 19
SKI	Shprintzen-Goldberg Craniosynostosis Syndrome, 1p36 Deletion	AD	99.66	39 of 39
	Syndrome			
SLC12A6	Corpus Callosum Agenesis-Neuronopathy Syndrome	AR	100	21 of 21
SLC25A24	Fontaine Progeroid Syndrome, Gorlin-Chaudhry-Moss Syndrome	AD	99.59	2 of 2
SLC2A10	Arterial Tortuosity Syndrome	AR	100	35 of 35
SLC35A2 SLC39A8	Congenital Disorder Of Glycosylation Congenital Disorder Of Glycosylation	X,XD,G AR	99.97 99.89	- 7 of 7
	, ,			128 of
SMAD3	Loeys-Dietz Syndrome, Aneurysm-Osteoarthritis Syndrome	AD	100	128
SMAD6	Craniosynostosis	AD	80.88	64 of 74
CNAC	Craniofacial Malformations With Polysyndactyly And Abnormal Skin And	AD	04.03	10 -f 10
SMO	Gut Development, Curry-Jones Syndrome	AR	94.03	10 of 10
SNX10	Autosomal Recessive Malignant Osteopetrosis	AR	100	14 of 14
SON	Zttk Syndrome, Brain Malformations-Musculoskeletal Abnormalities-	AD	99.27	30 of 32
	Facial Dysmorphism-Intellectual Disability Syndrome			
SOX10	Peripheral Demyelinating Neuropathy, Central Dysmyelination,	AD	99.74	139 of 147
SPECC1L	Waardenburg Syndrome Facial Clefting, Hypertelorism, Opitz Gbbb Syndrome	AD	99.66	147 14 of 14
	Hyper-IgE Recurrent Infection Syndrome, Permanent Neonatal Diabetes			171 of
STAT3	Mellitus	AD	100	171
	Intellectual Developmental Disorder With Autistic Features And Language			
TANC2	Delay, With Or Without Seizures, Non-Specific Syndromic Intellectual	AD	97.81	21 of 21
	Disability			
TARS1	Nonphotosensitive Trichothiodystrophy	AR	99.94	-
TBC1D24	Doors Syndrome, Rolandic Epilepsy With Paroxysmal Exercise-Induced	AD,AR	100	80 of 80
TBX1	Dystonia Andwriter's Cramp, Myoclonic Epilepsy DiGeorge Syndrome, Velocardiofacial Syndrome	AD,AR	88.7	35 of 42
TCF12	Craniosynostosis, Isolated Brachycephaly, Isolated Plagiocephaly	AD,AR	99.98	73 of 76
				140 of
TCIRG1	Osteopetrosis, Dysosteosclerosis	AR	100	146
TCOF1	Treacher Collins-Franceschetti Syndrome	AD	100	326 of
	·			327
TGFB2	Loeys-Dietz Syndrome	AD	99.9	41 of 44
TGFB3	Loeys-Dietz Syndrome	AD	100	34 of 35
TGFBR1	Loeys-Dietz Syndrome	AD	94	96 of 100
TGFBR2	Loeys-Dietz Syndrome	AD	99.9	165 of 166
TLK2	Autosomal Dominant Mental Retardation	AD	96.98	39 of 39
TMCO1	Cerebrofaciothoracic Dysplasia	AR	88	5 of 5
TNFSF11	Autosomal Recessive Malignant Osteopetrosis	AR	99.84	4 of 4
TRAIP	Seckel Syndrome	AR	100	2 of 2
	Craniosynostosis, Robinow-Sorauf Syndrome, Saethre-Chotzen			133 of
TWIST1	Syndrome, Sweeney-Cox Syndrome, Isolated Brachycephaly, Isolated	AD	74.06	161
	Plagiocephaly, Isolated Scaphocephaly		65.5	
TWIST2	Ablepharon-Macrostomia Syndrome, Barber-Say Syndrome	AD,AR	99.82	9 of 9
UFD1	22q11.2 Deletion Syndrome	- A D	99.98	47 of 40
WDR19	Cranioectodermal Dysplasia, Senior-Loken Syndrome, Jeune Syndrome	AR	99.96	47 of 49





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

<sup>\*</sup>Inheritance: AD: Autosomal Dominant; AR: Autosomal Recessive; X: X linked; XLR: X linked Recessive; Mi: Mitochondrial; Mu: Multifactorial.

# Methodology





#### 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.

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<sup>\*\*</sup>Number of clinically relevant mutations according to  $\mathsf{HGMD}$