Omphalocele and Gastroschisis

Precision Panel

Overview

Omphalocele, also known as exomphalos, is a midline abdominal wall defect at the base of the umbilical cord where herniation of abdominal contents takes place. The herniated organs are covered by the parietal peritoneum. The cause of omphalocele postulated to be a failure of the bowel to return into the abdomen by 10-12 weeks. Omphaloceles are associated with other anomalies in more than 70% of the cases, generally chromosomal, and the severity is dictated by the anomalies that are present. The main difficulty of this condition is the exclusion of associated conditions, not all diagnosed prenatally. Gastroschisis represents a herniation of abdominal contents through a paramedian full-thickness abdominal fusion defect. The abdominal herniation, in contrast with omphalocele, is usually to the right of the umbilical cord. It usually contains small bowel and has no surrounding membrane. Challenges in management of gastroschisis are related to the prevention of late intrauterine death, and the prediction and treatment of complex forms.

The Igenomix Omphalocele and Gastroschisis Gene Panel can be used to as a screening tool for underlying genetic alterations associated to these conditions. 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 Omphalocele and Gastroschisis Gene Panel is indicated for those patients with a clinical suspicion of omphalocele and/or gastroschisis which manifest as:

- Herniation of intestines through abdominal wall
- Polyhydramnios in utero
- Elevated levels of maternal serum a-fetoprotein (MSAFP)

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 with a multidisciplinary team for caesarean delivery, surgical repair and management of underlying associated conditions to prevent complications and ultimately lead to a better prognosis of the disease.
- Risk assessment of asymptomatic family members according to the mode of inheritance.

Genes & Diseases

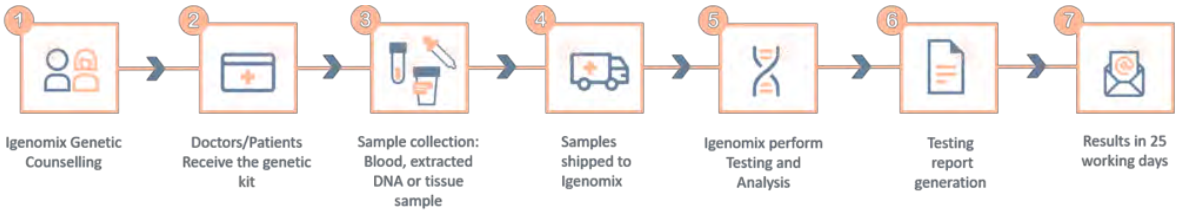
GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ACTG2	Familial Visceral Myopathy, Megacystis-Microcolon-Intestinal Hypoperistalsis Syndrome	AD	99.91	23 of 23
ALG9	Congenital Disorder Of Glycosylation Type II, Polycystic Kidney Disease Potter Type I With Microbrachycephaly, Hypertelorism And Brachymelia	AR	99.99	6 of 6
AMER1	Osteopathia Striata With Cranial Sclerosis	X,XD,G	99.45	NA of NA
BHLHA9	Camptosynpolydactyly, Gollop-Wolfgang Complex, Mesoaxial Synostotic Syndactyly With Phalangeal Reduction, Tibial Aplasia-Ectrodactyly Syndrome	AR	43.88	6 of 7
CD96	C Syndrome	AD	100	4 of 4
CDKN1C	Beckwith-Wiedemann Syndrome, Intrauterine Growth Retardation, Metaphyseal Dysplasia, Adrenal Hypoplasia Congenita And Genital Anomalies, Image Syndrome	AD	73.58	55 of 76
CEP120	Joubert Syndrome, Short-Rib Thoracic Dysplasia With Or Without Polydactyly, Jeune Syndrome, Joubert Syndrome	AR	99.8	9 of 9
CHD7	Charge Syndrome, Hypogonadotropic Hypogonadism With Or Without Anosmia, Kallmann Syndrome, Omenn Syndrome	AD	96.25	823 of 896
CHUK	Cocoon Syndrome	AR	100	5 of 5
COL11A1	Autosomal Dominant Deafness, Fibrochondrogenesis, Marshall Syndrome, Stickler Syndrome Type II, Autosomal Dominant Myopia-Midfacial Retrusion-Sensorineural Hearing Loss-Rhizomelic Dysplasia Syndrome	AD,AR	100	104 of 106
COL11A2	Autosomal Dominant Deafness, Fibrochondrogenesis, Otospondylomegaepiphyseal Dysplasia, Stickler Syndrome Type III	AD,AR	99.98	58 of 58
DACT1	Townes-Brocks Syndrome, Craniorachischisis	AD	98.12	8 of 9
DHCR7	Smith-Lemli-Opitz Syndrome	AR	100	217 of 217
DVL3	Autosomal Dominant Robinow Syndrome	AD	100	16 of 16
DYNC2H1	Short-Rib Thoracic Dysplasia With Or Without Polydactyly, Jeune Syndrome, Verma-Naumoff Type	AR,MU,D	99.78	214 of 221
DYNC2I1	Short-Rib Thoracic Dysplasia With Or Without Polydactyly, Jeune Syndrome, Verma-Naumoff Type	AR	97.76	14 of 14
DYNC2I2	Short-Rib Thoracic Dysplasia With Or Without Polydactyly, Jeune Syndrome, Verma-Naumoff Type	AR	99.54	23 of 23
FGFR1	Encephalocraniocutaneous Lipomatosis, Hartsfield Syndrome, Jackson-Weiss Syndrome, Kallmann Syndrome, Osteoglophonic Dysplasia, Pfeiffer Syndrome, Trigonocephaly, Lobar Holoprosencephaly, Microform Holoprosencephaly, Normosmic Congenital Hypogonadotropic Hypogonadism, Oligodontia, Septo-Optic Dysplasia Spectrum	AD	100	279 of 280
FLNA	X-Linked Cardiac Valvular Dysplasia, Frontometaphyseal Dysplasia, Periventricular Heterotopia, Chronic Neuronal Intestinal Pseudoobstruction, Melnick-Needles Syndrome, Otopalatodigital Syndrome, Terminal Osseous Dysplasia, Congenital Short Bowel Syndrome, X-linked Ehlers-Danlos Syndrome	X,XR,XD,G	100	NA of NA
FLNB	Atelosteogenesis, Boomerang Dysplasia, Larsen Syndrome, Spondylocarpotarsal Synostosis Syndrome	AD,AR	100	124 of 124
FOXF1	Alveolar Capillary Dysplasia With Misalignment Of Pulmonary Veins, Congenital Alveolar Capillary Dysplasia	AD	95.93	74 of 96
FRAS1	Fraser Syndrome	AR	98.73	57 of 58
FREM1	Bifid Nose With Or Without Anorectal And Renal Anomalies, Manitoba Oculotrichoanal Syndrome, Trigonocephaly, Bnar Syndrome, Isolated Trigonocephaly, Oculotrichoanal Syndrome	AD,AR	97.32	27 of 30
FREM2	Unilateral Or Bilateral Cryptophthalmos, Fraser Syndrome	AR	99.92	31 of 33
GPC3	Simpson-Golabi-Behmel Syndrome, Wilms Tumor, Nephroblastoma	AD,X,XR,G	99.84	NA of NA
GPC4	Keipert Syndrome, Simpson-Golabi-Behmel Syndrome	AD,X,XR,G	98.43	NA of NA
GRIP1	Fraser Syndrome	AR	100	17 of 17
H19-ICR	Beckwith-Wiedemann Syndrome, Multiple Tumor-Associated Chromosome Region, Silver-Russell Syndrome	AD	na	na
HIC1	Miller-Dieker Syndrome		97.7	NA of NA

HOXD13	Brachydactyly-Syndactyly Syndrome, Vacterl/Vater Association	AD	90.98	21 of 31
HYLS1	Hydrolethalus Syndrome, Joubert Syndrome	AR	100	2 of 2
IFT80	Asphyxiating Thoracic Dystrophy, Jeune Syndrome, Short Rib-Polydactyly Syndrome, Verma-Naumoff Type	AR	99.96	16 of 16
IFT81	Short-Rib Thoracic Dysplasia With Or Without Polydactyly	AR	98.97	7 of 9
IGF2	Beckwith-Wiedemann Syndrome, Silver-Russell Syndrome, Wilms Tumor, Isolated Hemihyperplasia	AD,X,XR,G	100	9 of 9
ISL1	Bladder Exstrophy		100	8 of 10
KCNQ1	Familial Atrial Fibrillation, Beckwith-Wiedemann Syndrome, Jervell And Lange-Nielsen Syndrome, Long QT Syndrome, Short QT Syndrome, Romano-Ward Syndrome	AD,AR	93.23	600 of 624
KCNQ1OT1	Beckwith-Wiedemann Syndrome, Isolated Hemihyperplasia	AD	na	na
LBR	Hydrops-Ectopic Calcification-Moth-Eaten Skeletal Dysplasia, Pelger-Huet Anomaly, Reynolds Syndrome, Greenberg Dysplasia	AD,AR	99.98	34 of 34
LMOD1	Megacystis-Microcolon-Intestinal Hypoperistalsis Syndrome		99.62	1 of 1
LONP1	Codas Syndrome	AR	99.84	21 of 21
LRP2	Donnai-Barrow Syndrome	AR	99.99	58 of 58
MASP1	3mc Syndrome	AR	100	29 of 30
MBTPS2	Ichthyosis Follicularis Atrichia And Photophobia Syndrome, Keratosis Follicularis Spinulosa Decalvans, Osteogenesis Imperfecta Type XIX, Palmoplantar Keratoderma, Bresek Syndrome, Ichthyosis Follicularis-Alopecia-photophobia Syndrome Mutilating Palmoplantar Keratoderma With Periorificial Keratotic Plaques	X,XR,G	100	NA of NA
MKS1	Bardet-Biedl Syndrome, Joubert Syndrome, Meckel Syndrome	AR	99.98	49 of 49
MMP14	Winchester Syndrome, Multicentric Osteolysis-Nodulosis-Arthropathy Spectrum		99.91	6 of 6
MMP2	Multicentric Osteolysis Nodulosis And Arthropathy Spectrum	AR	100	24 of 24
MTHFR	Homocystinuria Due To Deficiency Of N(5,10)-Methylene Tetrahydrofolate Reductase Activity , Neural Tube Defects, Thrombophilia, Isolated Anencephaly, Isolated Exencephaly	AD,AR	100	122 of 122
MYH11	Familial Thoracic Aortic Aneurysm, Familial Aortic Dissection, Megacystis-Microcolon-Intestinal Hypoperistalsis Syndrome	AD	100	67 of 67
MYLK	Familial Thoracic Aortic Aneurysm, Megacystis-Microcolon-Intestinal Hypoperistalsis Syndrome	AD	99.95	50 of 50
NEK9	Arthrogryposis, Perthes Disease And Upward Gaze Palsy, Lethal Congenital Contracture Syndrome, Nevus Comedonicus Syndrome	AR	99.98	4 of 4
NFIX	Marshall-Smith Syndrome, Sotos Syndrome, 19p13.3 Microduplication Syndrome, Malan Overgrowth Syndrome, Marshall-Smith Syndrome	AD	94.42	75 of 81
NXN	Autosomal Recessive Robinow Syndrome	AR	98.03	2 of 4
PAFAH1B1	Lissencephaly, 17p13.3 Microduplication Syndrome, Miller-Dieker Syndrome	AD	99.95	90 of 92
PIGN	Multiple Congenital Anomalies-Hypotonia-Seizures Syndrome, Fryns Syndrome	AR	93.97	36 of 39
PIGY	Hyperphosphatasia With Mental Retardation Syndrome, Hyperphosphatasia-Intellectual Disability Syndrome	AR	100	1 of 2
PORCN	Focal Dermal Hypoplasia	X,XD,G	100	NA of NA
PPP1R12A	Genitourinary And/Or/Brain Malformation Syndrome	AD	99.48	1 of 1
PPP2R3C	Gonadal Dysgenesis, Dysmorphic Facies, Retinal Dystrophy, And Myopathy, Spermatogenic Failure	AD,AR	99.85	3 of 3
PTCH1	Basal Cell Nevus Syndrome, Holoprosencephaly, Alobar Holoprosencephaly, Gorlin Syndrome, Lobar Holoprosencephaly, Microform Holoprosencephaly, Midline Interhemispheric Variant Of Holoprosencephaly, Monosomy 9q22.3, Semilobar Holoprosencephaly, Septopreoptic Holoprosencephaly	AD	98.89	498 of 502
RAB23	Carpenter Syndrome	AR	100	15 of 15
SEMA3E	Charge Syndrome, Hypogonadotropic Hypogonadism Without Anosmia	AD,AR	99.81	6 of 7
SF3B4	Acrofacial Dysostosis, Nager Syndrome	AD	94.86	33 of 40
SPECC1L	Facial Clefting, Oblique, Hypertelorism, Opitz Gbbb Syndrome, Autosomal Dominant	AD	99.66	14 of 14
THRA	Congenital Nongoitrous Hypothyroidism	AD	100	24 of 24
TP63	Ankyloblepharon-Ectodermal Defects-Cleft Lip/Palate, Ectrodactyly, Ectodermal Dysplasia, And Cleft Lip/Palate Syndrome, Limb-Mammary Syndrome, Rapp-Hodgkin Syndrome, Split-Hand/Foot Malformation, Adult Syndrome, Palate Syndrome, Bladder Exstrophy	AD	99.98	144 of 144
TSHB	Congenital Nongoitrous Hypothyroidism, Isolated Thyroid-stimulating Hormone Deficiency	AR	100	12 of 13

TTC7A	Gastrointestinal Defects And Immunodeficiency Syndrome, Combined Immunodeficiency-Enteropathy Spectrum, Multiple Intestinal Atresia	AR	100	44 of 45
TWIST2	Ablepharon-Macrostomia Syndrome, Barber-Say Syndrome, Focal Facial Dermal Dysplasia	AD,AR	99.82	9 of 9
VANGL2	Neural Tube Defects, Isolated Anencephaly, Isolated Exencephaly	AD	99.98	12 of 12
WDR35	Cranioectodermal Dysplasia, Short-Rib Thoracic Dysplasia With Or Without Polydactyly, Cranioectodermal Dysplasia, Short Rib-Polydactyly Syndrome, Verma-Naumoff Type	AR	100	31 of 33
WNT3	Tetraamelia-Multiple Malformations Syndrome	AR	100	2 of 2
YWHAE	17p13.3 Microduplication Syndrome, Miller-Dieker Syndrome		98.99	0 of 1
ZIC3	X-linked Visceral Heterotaxy, Vacterl Association With Hydrocephalus	X,XR,G	99.98	NA of NA

*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



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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

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