Primary Microcephaly Precision Panel

Overview

Microcephaly is generally defined as a small head size, typically greater than two standard deviations below normal, as measured via occipital frontal circumference, using a measuring tape. Microcephaly is a clinical sign and not a disease. It is a result of a premature fusion of the skull sutures that leads to an abnormal shape and a growth limitation of the head circumference that can later result in long term neurological sequelae. The cause of microcephaly can be divided in two groups: premature fusion of cranial sutures, also known as craniosynostosis or poor brain growth. Genetic conditions are one of the etiologies of microcephaly and include: Trisomy 21, 13, 18, Cri Du Chat syndrome, Williams syndrome etc. Also, inborn errors of metabolism, disruptive injuries and maternal deprivation problems can lead to microcephaly. Primary microcephaly is usually inherited in an autosomal recessive pattern.

The Igenomix Primary Microcephaly Precision Panel can be as a diagnostic tool to reveal underlying genetic conditions 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, and their high or intermediate penetrance.

Indications

The Igenomix Primary Microcephaly Precision Panel is indicated in those cases where there is:

- Head circumference more than 2 standard deviations below the mean
- Consistent low percentile of head circumference ultrasound measurements during pregnancy
- Associated syndromic features: congenital heart defects, umbilical hernia, hypotonia, low set ears, short neck, overlapping fingers, micrognathia, polydactyly etc

Clinical Utility

The clinical utility of this panel is:

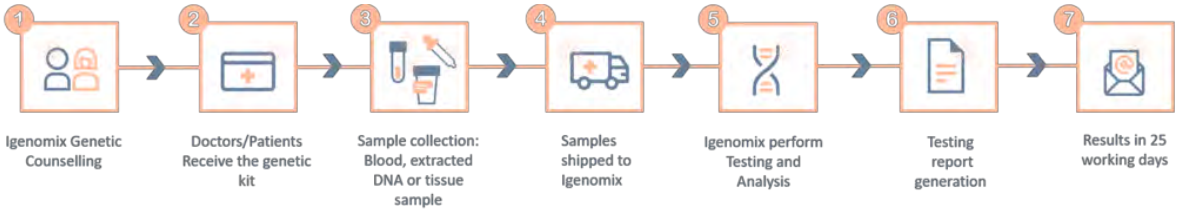
- The genetic and molecular diagnosis for an accurate clinical diagnosis of microcephaly as well as underlying genetic conditions.
- Early initiation of treatment with a multidisciplinary team for appropriate surveillance, surgical care and long-term monitoring to ensure proper cerebral and cranial growth.
- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.
- Improved pathways from diagnosis to treatment in susceptible populations.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ANKLE2	Autosomal Recessive Primary Microcephaly	AR	96.08	4 of 4
ASPM	Autosomal Recessive Primary Microcephaly	AR	99.74	221 of 222
CDK5RAP2	Autosomal Recessive Primary Microcephaly	AR	100	32 of 32
CDK6	Autosomal Recessive Primary Microcephaly	AR	100	1 of 1
CENPE	Autosomal Recessive Primary Microcephaly, Seckel Syndrome	AR	95.69	5 of 5
CENPJ	Autosomal Recessive Primary Microcephaly, Seckel Syndrome	AR	99.97	13 of 13
CEP135	Autosomal Recessive Primary Microcephaly	AR	99.48	7 of 8
CEP152	Autosomal Recessive Primary Microcephaly, Seckel Syndrome	AR	97.73	21 of 24
CEP63	Autosomal Recessive Primary Microcephaly, Seckel Syndrome	AR	100	3 of 3
CIT	Autosomal Recessive Primary Microcephaly	AR	99.98	17 of 17
COPB2	Autosomal Recessive Primary Microcephaly	AR	99.64	4 of 4
DPP6	Autosomal Dominant Primary Microcephaly, Paroxysmal Familial Ventricular Fibrillation, Autosomal Dominant Mental Retardation	AD	97.03	23 of 28
KIF14	Autosomal Recessive Primary Microcephaly, Meckel Syndrome	AR	99.84	18 of 18
KNL1	Autosomal Recessive Primary Microcephaly	AR	98.91	NA of NA
TRAPPC14	Autosomal Recessive Primary Microcephaly	AR	na	na
MCPH1	Autosomal Recessive Primary Microcephaly	AR	99.51	18 of 19
MFSD2A	Autosomal Recessive Primary Microcephaly	AR	97.58	6 of 6
NCAPD2	Autosomal Recessive Primary Microcephaly	AR	99.98	4 of 4
NCAPD3	Autosomal Recessive Primary Microcephaly	AR	99.97	4 of 5
NCAPH	Autosomal Recessive Primary Microcephaly	AR	99.99	1 of 1
NUP37	Autosomal Recessive Primary Microcephaly	AR	100	3 of 3
PHC1	Autosomal Recessive Primary Microcephaly	AR	91.73	1 of 1
PPP1R15B	Primary Microcephaly-Mild Intellectual Disability-Young-Onset Diabetes Syndrome	AR	99.98	5 of 5
PYCR2	Autosomal Recessive Primary Microcephaly , Pycr2-Related Microcephaly-Progressive Leukoencephalopathy	AR	98.29	14 of 14
SASS6	Autosomal Recessive Primary Microcephaly	AR	99.14	6 of 6
STIL	Autosomal Recessive Primary Microcephaly	AR	99.94	18 of 18
TAF13	Autosomal Recessive Primary Microcephaly	AR	99.97	5 of 5
TRMT10A	Primary Microcephaly-Mild Intellectual Disability-Young-Onset Diabetes Syndrome	AR	99.81	7 of 7
WDFY3	Autosomal Dominant Primary Microcephaly, Non-Specific Syndromic Intellectual Disability	AD	99.95	60 of 60
WDR62	Autosomal Recessive Primary Microcephaly With Or Without Cortical Malformations	AR	100	60 of 61
ZNF335	Microcephalic Primordial Dwarfism Due To Znf335 Deficiency, Autosomal Recessive Primary Microcephaly	AR	99.83	20 of 20

*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

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