Aicardi-Goutieres Syndrome

Precision Panel

Overview

Aicardi-Goutieres Syndrome is a rare genetic neurological disorder with variable clinical manifestations including infantile spasms-in-flexion, total or partial agenesis of the corpus callosum, and variable ocular abnormalities. It usually manifests as early-onset encephalopathy resulting in severe intellectual and physical disability. Genetic diagnosis is crucial due to the overlapping phenotypic features with sequelae of congenital infection and systemic lupus erythematosus (SLE). Given the phenotypic heterogeneity and diagnostic difficulties associated with young children, Aicardi-Goutieres syndrome may be a more frequent cause of mental retardation and seizure in girls than previously thought. Complications of this syndrome include severe mental retardation, intractable epilepsy and pulmonary complications. It is typically inherited in an autosomal recessive pattern.

The Igenomix Aicardi-Goutieres Precision Panel can serve as an accurate and directed diagnostic tool as well as a differential diagnosis for early-onset encephalopathy 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 Aicardi-Goutieres Syndrome Precision Panel is indicated in patients with a clinical suspicion or diagnosis with or without the following manifestations:

- Seizures
- Ocular abnormalities
- Global developmental delay
- Inability to walk due to spastic hemiplegia
- Early presentation of malignancies such as soft tissue carcinoma, hepatoblastoma and angiosarcoma
- Unusually small head
- Hand deformities
- Gastrointestinal difficulties: diarrhea, constipation, difficulty eating etc

Clinical Utility

The clinical utility of this panel is:

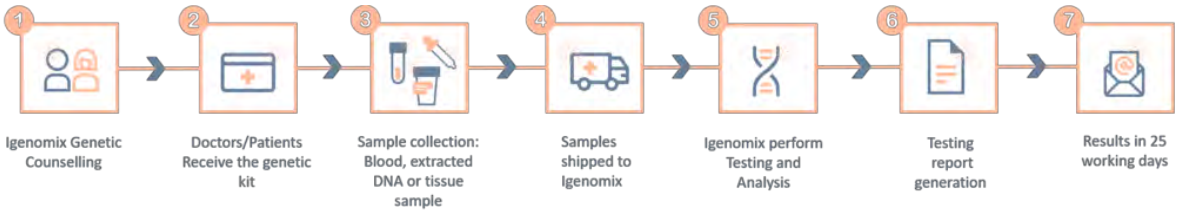
- The genetic and molecular diagnosis for an accurate clinical diagnosis of a symptomatic patient.
- Early initiation of treatment with a multidisciplinary team in the form of medical care of seizures, physical medicine and rehabilitation, prevention of pulmonary complications and early surveillance for neoplasms. Surgical care is indicated for refractory intractable epilepsy.
- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.
- Improve correlation of genotype and phenotype to increase diagnostic accuracy.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ADAR	Aicardi-Goutieres Syndrome, Dyschromatosis Symmetrica Hereditaria, Familial Infantile Bilateral Striatal Necrosis	AD,AR	99.93	252 of 252
IFIH1	Aicardi-Goutieres Syndrome, Singleton-Merten Syndrome	AD	99.62	26 of 27
RNASEH2A	Aicardi-Goutieres Syndrome	AR	100	23 of 23
RNASEH2B	Aicardi-Goutieres Syndrome	AR	99.95	41 of 41
RNASEH2C	Aicardi-Goutieres Syndrome	AR	100	14 of 14
SAMHD1	Aicardi-Goutieres Syndrome, Chilblain Lupus	AD,AR	100	51 of 51
TREX1	Aicardi-goutieres Syndrome, Chilblain Lupus, Systemic Lupus Erythematosus, Vasculopathy, Retinal, With Cerebral Leukodystrophy	AD,AR	100	75 of 75

*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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