Ataxia Telangiectasia

Precision Panel

Overview

Ataxia Telangiectasia is a complex multisystem disorder characterized by progressive neurologic impairment, altered balance, variable immunodeficiency with susceptibility to upper respiratory tract infections, impaired organ maturation, ocular and cutaneous telangiectasia and predisposition to malignancy. Malignancy usually involves the lymphatic system (i.e., lymphomas) and blood-forming organs (i.e., leukemia) as well as the brain. It is a primary immunodeficiency of B and T cells caused by a mutation in genes encoding DNA repair enzymes. This disease has a very heterogeneous presentation and genetic background, as shown by the existence of 4 complementation groups (A,C,D,E). As well, there is a variable rate of progression leading to death often by early adulthood. It is inherited in an autosomal recessive manner.

The Igenomix Ataxia Telangiectasia Precision Panel can be as a tool for an accurate diagnosis as well as differential 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, and their high or intermediate penetrance.

Indications

The Igenomix Ataxia Telangiectasia Precision Panel is used for patients with a clinical suspicion or diagnosis of Ataxia Telangiectasia presenting with the following symptoms:

- Ocular and cutaneous telangiectasia (tiny, red, spider-like veins)
- Decreased coordination of movements (ataxia)
- Abnormal eye movements
- Abnormal head movements
- Cerebral palsy
- Developmental delay
- Growth retardation
- Recurrent respiratory infections

Clinical Utility

The clinical utility of this panel is:

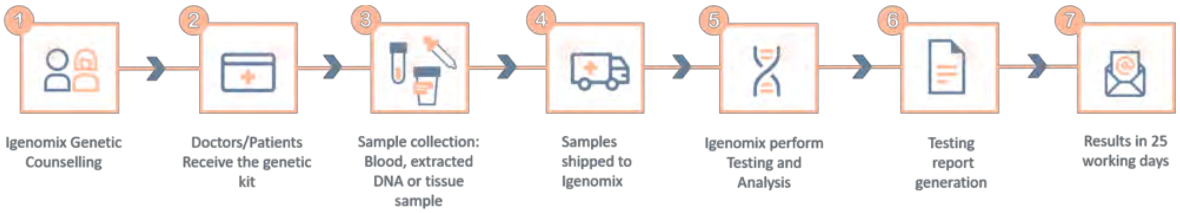
- The genetic and molecular confirmation for an accurate clinical diagnosis of a symptomatic patient.
- Early initiation of treatment involving a multidisciplinary team focusing on preventive care of infections and other complications, symptomatic medical care for neurologic symptoms alongside early surveillance for cancer detection.
- Risk assessment of asymptomatic family members according to the mode of inheritance via genetic counselling and explanation of the multisystem nature of the disease.
- Improvement of delineation of genotype-phenotype correlation.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ATM	Ataxia-Telangiectasia, Breast Cancer, Mantle Cell Lymphoma	AD,AR	99.93	1608 of 1632
MRE11	Ataxia-Telangiectasia-like Disorder, Hereditary Breast And Ovarian Cancer Syndrome	AR	99.95	NA of NA
NBN	Aplastic Anemia, Acute Lymphocytic Leukemia, Nijmegen Breakage Syndrome, Hereditary Breast And Ovarian Cancer Syndrome	AR,MU,P	100	200 of 200
PCNA	Ataxia-Telangiectasia-like Disorder, PCNA-Related Progressive Neurodegenerative Photosensitivity Syndrome	AR	99.92	1 of 1

*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

1. Soresina, A., Meini, A., Lougaris, V., Cattaneo, G., Pellegrino, S., & Piane, M. et al. (2008). Different Clinical and Immunological Presentation of Ataxia-Telangiectasia within the Same Family. *Neuropediatrics*, *39*(1), 43-45. doi: 10.1055/s-2008-1076736
2. McGrath-Morrow, S. A., Ndeh, R., Helmin, K. A., Khuder, B., Rothblum-Oviatt, C., Collaco, J. M., Wright, J., Reyfman, P. A., Lederman, H. M., & Singer, B. D. (2020). DNA methylation and gene expression signatures are associated with ataxia-telangiectasia phenotype. *Scientific reports*, *10*(1), 7479. https://doi.org/10.1038/s41598-020-64514-2
3. van Os, N., Chessa, L., Weemaes, C., van Deuren, M., Fiévet, A., van Gaalen, J., Mahlaoui, N., Roeleveld, N., Schrader, C., Schindler, D., Taylor, A., Van de Warrenburg, B., Dörk, T., & Willemsen, M. (2019). Genotype-phenotype correlations in ataxia telangiectasia patients with *ATM* c.3576G>A and c.8147T>C mutations. *Journal of medical genetics*, *56*(5), 308–316. https://doi.org/10.1136/jmedgenet-2018-105635

4. Verhagen, M. (2011). 1FC3.3 The amounts of ATM protein and residual kinase activity predict the phenotype in Ataxia-Telangiectasia: A genotype – phenotype study. *European Journal Of Paediatric Neurology*, *15*, S15. doi: 10.1016/s1090-3798(11)70055-1
5. Liu, X., Wang, T., Huang, X., Zhou, H., Luan, X., & Shen, J. et al. (2016). Novel ATM mutations with ataxia-telangiectasia. *Neuroscience Letters*, *611*, 112-115. doi: 10.1016/j.neulet.2015.11.036