alamya health

A Future Where Precision Medicine is the Standard

Repeat Alteration Disorders Screening

Alamya Health, PBC
411 Lake Branch Road
Clever, MO 65631 USA

Alamya Health, PTE Ltd
No. 8 Burn Road
#04-04 Trivex
Singapore 369977

Repeat Alteration Disorders Screening

Background

Many genes contain repeated short sequences of DNA. The number of repeats can vary among healthy individuals and within families. Repeat alteration disorders (e.g., trinucleotide repeat disorders) are genetic conditions caused by the number of repeat units being outside the range seen in healthy individuals. Some altered repeat sequences can interfere with normal gene expression and protein function and lead to progressive neurological, muscular, or developmental disorders. The severity and onset of symptoms typically depend on the number of repeat units, which can change across generations. Examples of such disorders include Huntington disease, fragile X syndrome, and myotonic dystrophy.

Most of these disorders do not have effective preventions or treatments. The decision to pursue repeat alteration disorder screening is entirely up to you. You may wish to discuss this with your healthcare professional.

The table below lists the genes that are screened and the names of the disorders associated with pathogenic repeat alterations in these genes. The Online Mendelian Inheritance in Man (OMIM) number for each disorder is also listed. OMIM is an online encyclopedia of genetic disorders accessible at OMIM.

The Alamya Health team updates the gene list based on current scientific knowledge. Relevant resources and references are provided for positive test findings.

Limitations

Repeat alteration screening identifies most people who have repeat alterations for the conditions screened. The interpretation of the testing results relies on the current knowledge of the genetic basis of these disorders. As this knowledge is constantly changing, not all pathogenic repeat alteration disorders are currently known. Additionally, limitations of the sequencing technology and interpretation parameters can fail to detect repeat alterations that could lead to disease.

The term screening is used because not all repeat alterations are screened, and not all repeat alterations can be identified by the testing technology. A ‘negative’ result greatly reduces the risk to develop one of the repeat alteration disorders screened; it does not completely rule them out. Age, clinical history, and family history also influence risk. .

REPEAT ALTERATION DISORDERS SCREENED

Gene Symbol	Disease	OMIM Number
AFF2	Fragile X syndrome, FRAXE type	309548
AR	Spinal and bulbar muscular atrophy of Kennedy	131200
ATN1	Dentatorubral-pallidoluysian atrophy	125370
ATXN1	Spinocerebellar ataxia 1	164400
ATXN10	Spinocerebellar ataxia 10	603516
ATXN2	Spinocerebellar ataxia 2	183090
ATXN3	Machado-Joseph disease	109150
ATXN7	Spinocerebellar ataxia 7	164500
ATXN8OS	Spinocerebellar ataxia 8	608768
C9ORF72	Frontotemporal dementia and/or amyotrophic lateral sclerosis 1	105550
CACNA1A	Spinocerebellar ataxia 6	183086
CBL	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia	613563
CNBP	Myotonic dystrophy 2	602668
CSTB	Progressive myoclonic epilepsy 1A	254800
DIP2B	Intellectual developmental disorder, autosomal dominant, FRA12A type	136630
DMPK	Myotonic dystrophy 1	160900
FMR1	Fragile X syndrome	300624
	Fragile X tremor/ataxia syndrome	300623
	Premature ovarian failure 1	311360
FXN	Friedreich ataxia	229300
GLS	Global developmental delay, progressive ataxia, and elevated glutamine	618412
HTT	Huntington disease	143100
JPH3	Huntington disease-like 2	606438
NIPA1	Spastic paraplegia 6	600363
NOP56	Spinocerebellar ataxia 36	614153
NOTCH2NLC	Neuronal intranuclear inclusion disease	603472
PABPN1	Oculopharyngeal muscular dystrophy	164300
PHOX2B	Congenital central hypoventilation syndrome	209880
PPP2R2B	Spinocerebellar ataxia 12	604326
RFC1	RFC1 CANVAS / spectrum disorder	614575
TBP	Spinocerebellar ataxia type 17	607136
TCF4	Fuchs endothelial corneal dystrophy-3	613267