Genomic Unity® Testing

Tandem repeats analyzed

The following characterized repeats are detected and analyzed by the Genomic Unity® test.

Gene(s)	Repeat	Disorder
AFF2	CCG	Fragile XE syndrome
AR	CAG	Spinal and bulbar muscular atrophy
ATN1	CAG	Dentatorubral-pallidoluysian atrophy (DRPLA)
ATXN1	CAG	Spinocerebellar ataxia
ATXN10	ATTCT	Spinocerebellar ataxia
ATXN2	CAG	Spinocerebellar ataxia
ATXN3	CAG	Spinocerebellar ataxia
ATXN7	CAG	Spinocerebellar ataxia
ATXN80S	CTG	Spinocerebellar ataxia
C9ORF72	GGGGCC	Frontotemporal dementia and/or amyotrophic lateral sclerosis (FTDALS1)
CACNA1A	CAG	Spinocerebellar ataxia
CNBP	CCTG	Myotonic dystrophy type II
CSTB	CCCCGCCCCGCG	Myoclonus epilepsy
DIP2B	CGG	FRA12A fragile site
DMPK	CTG	Myotonic dystrophy type I
FMR1	CGG	Fragile X syndrome
FXN	GAA	Friedreich's ataxia
HTT	CAG	Huntington disease
JPH3	CTG	Huntington disease-like 2 syndrome
NOP56	GGCCTG	Spinocerebellar ataxia
NOTCH2NLC	GGC	Neuronal intranuclear inclusion disease
PHOX2B	Alanine	Congenital central hypoventilation syndrome
PABPN1	GCN	Oculopharyngeal muscular dystrophy
PPP2R2B	CAG	Spinocerebellar ataxia
TBP	Glutamine	Spinocerebellar ataxia
TCF4	CAG	Fuchs endothelial corneal dystrophy

Sensitivity and reporting policies differ by loci. The false negative rate for repeat expansions has not been determined for the following genes: *AFF2, ATXN10, CNBP, CSTB, DIP2B, NOTCH2NLC, PHOX2B, TBP.*

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