Genomic Unity® Testing

Short tandem repeat genes included in Genomic Unity®

Gene(s)	Repeat	Disorder
AFF2	CCG	Fragile XE syndrome
AR	CAG	Spinal and bulbar muscular atrophy
ARX (2 tracts)*	CGC	X-linked Intellectual developmental disorder 29 and Partington syndrome
ATN1	CAG	Dentatorubral-pallidoluysian atrophy (DRPLA)
ATXN1	CAG	Spinocerebellar ataxia (SCA1)
ATXN10	ATTCT	Spinocerebellar ataxia (SCA10)
ATXN2	CAG	Spinocerebellar ataxia (SCA2)
ATXN3	CAG	Spinocerebellar ataxia (SCA3)
ATXN7	CAG	Spinocerebellar ataxia (SCA7)
ATXN80S	CTG	Spinocerebellar ataxia (SCA8)
C9ORF72	GGGGCC	Frontotemporal dementia and/or amyotrophic lateral sclerosis (FTDALS1)
CACNA1A	CAG	Spinocerebellar ataxia (SCA6)
CNBP	CCTG	Myotonic dystrophy type II
CSTB	CCCCGCCCCGCG	Myoclonus epilepsy
DIP2B	CGG	FRA12A fragile site
DMPK	CTG	Myotonic dystrophy type I
FGF14	GAA	Late-onset spinocerebellar ataxia-27B (SCA27B)
FOXL2	GCN	Blepharophimosis, ptosis, and epicanthus inversus syndrome type II (BPES II)
FMR1	CGG	Fragile X syndrome
FXN	GAA	Friedreich's ataxia
GIPC1	GGC	Oculopharyngodistal myopathy 2 (OPDM2)
GLS	GCA	Global developmental delay, progressive ataxia, and elevated glutamine (GDPAG)
HTT	CAG	Huntington disease
JPH3	CTG	Huntington disease-like 2 syndrome
LRP12	CGG	Oculopharyngodistal myopathy 1 (OPDM1)
NOP56	GGCCTG	Spinocerebellar ataxia (SCA36)
NOTCH2NLC	GGC	Neuronal intranuclear inclusion disease
PHOX2B	Alanine	Congenital central hypoventilation syndrome

Gene(s)	Repeat	Disorder
PABPN1	GCN	Oculopharyngeal muscular dystrophy
PPP2R2B	CAG	Spinocerebellar ataxia (SCA12)
RFC1	AAGGG and other pathogenic repeats	Cerebellar ataxia, neuropathy, and vestibular areflexia syndrome (CANVAS)
SOX3	GCN	X linked intellectual developmental disorder with isolated growth hormone deficiency X linked panhypopituitarism
TBP	Glutamine	Spinocerebellar ataxia (SCA17)
TCF4	CAG	Fuchs endothelial corneal dystrophy
VWA1	GGCGCGGAGC	Hereditary distal motor neuropathy with myopathic features (HMNMYO)
ZIC2	GCN	Holoprosencephaly 5
ZFHX3	GGC	Spinocerebellar ataxia (SCA4)

Short tandem repeat genes additionally included in Genomic Unity® 2.0

Gene(s)	Repeat	Disorder
BEAN1	TGGAA	Spinocerebellar Ataxia 31 (SCA31)
HOXA13	GCN	Hand-Foot-Genital Syndrome (HFGS)
PRDM12	GCC	Hereditary Sensory and Autonomic Neuropathy Type VIII (HSAN8) Midfacial Toddler Excoriation Syndrome (MiTES)
PRNP	24-base octapeptide PHGGGWGQ	Prion Diseases
RILPL1	CGG	Qculopharyngodistal Myopathy Type 4 (OPDM4)
RUNX2	CAG followed by GCN	Cleidocranial Dysplasia (CCD)
SAMD12	$(TTTTA)_7$ TTA $(TTTTA)_{13}$	Benign Adult Familial Myoclonic Epilepsy (BAFME)

Sensitivity and reporting policies differ by loci. The false negative rate for repeat expansions has not been determined.

**ARX* repeat expansions will be reported only in cases where the clinical symptoms of the patient include early onset seizures.

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