PerkinElmer
For the Better

PerkinElmer GENOMICS

TANDEM REPEAT SCREENING BY WHOLE GENOME SEQUENCING

Historically, detection of repeat expansions has only been possible with polymerase chain reaction–based assays or Southern blots, which are costly and time-consuming methods. With new technology, we can now screen STRs on NGS based assays, allowing WGS the potential to profile nearly all genetic variants simultaneously in a single assay.

Tandem repeat screening by Whole Genome Sequencing can be used to rule out select repeat conditions by identifying and characterizing non-expanded (normal) alleles in the proband. The limit of detection of an expanded allele size on NGS based methods is currently not well established. Thus, if an allele is expanded, depending on the size, further confirmatory diagnostic testing may be required to fully characterize the repeat size to provide a definitive diagnosis.

TANDEM REPEAT SCREENING BY WHOLE GENOME SEQUENCING

At PerkinElmer Genomics, we include the screening of 35 genes for short tandem repeats in our whole genome sequencing (WGS) test, listed below.

Gene	Condition
AFF2	Fragile X syndrome, FRAXE type
AR	Spinal bulbar muscular atrophy
ATN1	Dentatorubral-pallidoluysian atrophy
ATXN1	Spinocerebellar ataxia type 1
ATXN10	Spinocerebellar ataxia type 10
ATXN2	Spinocerebellar ataxia type 2, L-dopa responsive parkinsonism ALS type 13
ATXN3	Spinocerebellar ataxia type 3
ATXN7	Spinocerebellar ataxia type 7
ATXN8OS	Spinocerebellar ataxia type 8
C9orf72	Frontotemporal dementia, ALS type 1, hereditary ataxia
CACNA1A	Spinocerebellar ataxia type 6
CD40LG	X-linked hyper IgM syndrome
CNBP	Myotonic dystrophy type 2
COMP	Pseudo-achondroplasia
CSTB	Unverricht-Lundborg disease
DIP2B	Mental retardation type 12A
DMPK	Myotonic dystrophy type 1
FMR1	Fragile X syndrome, fragile X-associated tremor/ataxia syndrome
FOXL2	Blepharophimosis, ptosis, epicanthus inversus
FXN	Friedreich ataxia
HOXD13	Syndactyly, type V
IL11RA	Craniosynostosis and dental anomalies
MYH7	Laing distal myopathy
NOP56	Spinocerebellar ataxia type 36
PABPN1	Oculopharyngeal muscular dystrophy
PAX2	Renal coloboma syndrome
PHOX2B	Congenital central hypoventilation syndrome
PPP2R2B	Spinocerebellar ataxia type 12
RUNX2	Cleidocranial dysplasia
SOX3	Panhypopituitarism and intellectual disability with growth hormone deficiency
TBP	Spinocerebellar ataxia type 17
TBX1	Tetralogy of Fallot
TCF4	Fuchs endothelial corneal dystrophy
ZIC2	Holoprosencephaly type 5
ZIC3	VACTERL