

Multiplex PCR (polyQ)
Polyglutamin repeat disorders v1*

HGNC approved gene symbol	Transcript	OMIM gene ID	Test**	Disorder
ATXN1	NM_001128164	601556	PCR and fragment analysis	SCA17
ATN1	NM_001940	607462	PCR and fragment analysis	DRPLA
ATXN2	NM_001372574	601517	PCR and fragment analysis	SCA2
ATXN3	NM_004993	607047	PCR and fragment analysis	SCA3
CACNA1A	NM_001127222	601011	PCR and fragment analysis	SCA6
ATXN7	NM_001377405	607640	PCR and fragment analysis	SCA7
TBP	NM_003194	600075	PCR and fragment analysis	SCA17
C9ORF72	NM_018325	105550	outsourced to AmsterdamUMC	
HTT***	NM_002111.5	613004	PCR and fragment analysis	HD

* repeat ranges (normal, normal mutable, expanded) are provided in the report

** number of repeats is determined by PCR across the repeat followed by fragment analysis

*** this is tested in a separate experiment