

TABLE 1 | Features of trinucleotide expansion in humans

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[Mechanisms of trinucleotide repeat instability during human development](#)

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Disease	Sequence	Location	Parent of origin of expansion	Repeat number (normal)	Repeat number (pre-mutation)	Repeat number (disease)	Somatic instability
Diseases with coding TNRs							
DRPLA	CAG	<i>ATN1</i> (exon 5)	P	6–35	35–48	49–88	Yes
HD	CAG	<i>HTT</i> (exon 1)	P	6–29	29–37	38–180	Yes
OPMD	GCN	<i>PABPN1</i> (exon 1)	P and M	10	12–17	>11	None found in tissue tested (hypothalamus)
SCA1	CAG	<i>ATXN1</i> (exon 8)	P	6–39	40	41–83	Yes
SCA2	CAG	<i>ATXN2</i> (exon 1)	P	<31	31–32	32–200	Unknown
SCA3 (Machado–Joseph disease)	CAG	<i>ATXN3</i> (exon 8)	P	12–40	41–85	52–86	Unknown
SCA6	CAG	<i>CACNA1A</i> (exon 47)	P	<18	19	20–33	None found
SCA7	CAG	<i>ATXN7</i> (exon 3)	P	4–17	28–33	>36 to >460	Yes
SCA17	CAG	<i>TBP</i> (exon 3)	P > M	25–42	43–48	45–66	Yes
SMBA	CAG	<i>AR</i> (exon 1)	P	13–31	32–39	40	None found
Diseases with non-coding TNRs							
DM1	CTG	<i>DMPK</i> (3' UTR)	M	5–37	37–50	<50	Yes
DM2	CCTG	<i>CNBP</i> (intron 1)	Uncertain	<30	31–74	75–11,000	Yes
FRAX-E	GCC	<i>AFF2</i> (5' UTR)	M	4–39	40–200	>200	Unknown
FRDA	GAA	<i>FXN</i> (intron 1)	Recessive	5–30	31–100	70–1,000	Yes
FXS	CGG	<i>FMR1</i> (5' UTR)	M	6–50	55–200	200–4,000	Yes
HDL2	CTG	<i>JPH3</i> (exon 2A)	M	6–27	29–35	36–57	Unknown
SCA8	CTG	<i>ATXN8OS</i> (3' UTR)	M	15–34	34–89	89–250	Unknown
SCA10	ATTCT	<i>ATXN10</i> (intron 9)	M and P (smaller changes with M)	10–29	29–400	400–4,500	Yes
SCA12	CAG	<i>PPP2R2B</i> (5' UTR)	M and P (more unstable with P)	7–28	28–66	66–78	None found