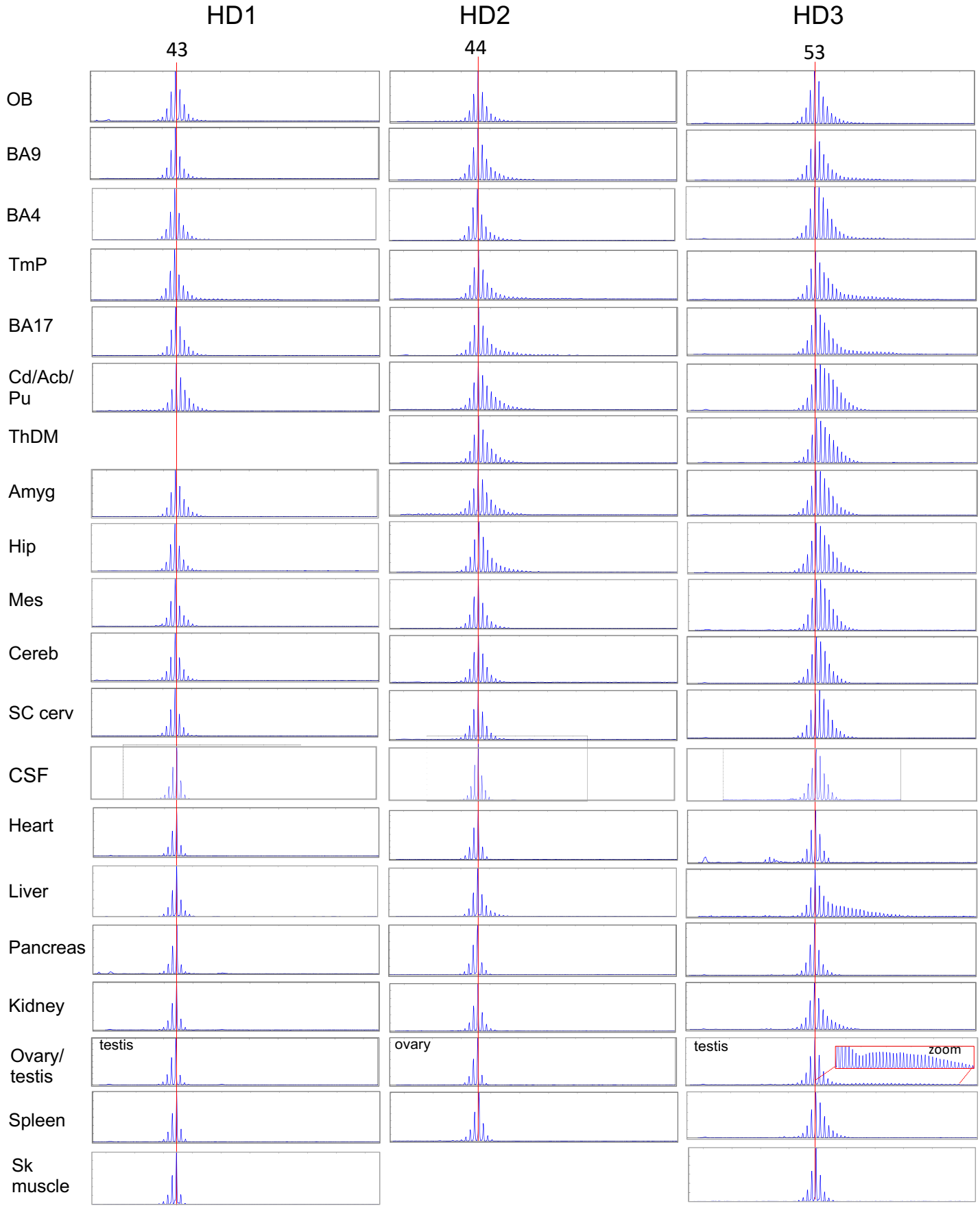
Table S1: Number of CNS and peripheral tissue samples analyzed in HD individuals

Brain areas	Abbreviation	Boston (USA)				Bochum (Germany)			
		HD1	HD2	HD3	HD4	HD5	HD6	HD7	HD8
Olfactory bulb	OB	3	3	3					
Cortex (straight gyrus, BA11)	BA11					1	1	1	
Cortex (prefrontal cortex, BA9)	BA9	3	3	3	2				
Cortex (anterior cingulate/midcingulate cortex, BA24)	BA24	3	3	3	3	1	1	1	3
Cortex (postcingulate cortex, BA23)	BA23					2	1	1	
Cortex (primary motor cortex, BA4)	BA4	3	3	3	3	1		1	
Cortex (primary somatosensory cortex, BA 3.1.2)	BA3.1.2	3	3	3	3				
Cortex (temporal pole)	TmP	3	3	3	2				
Cortex (primary visual cortex, BA17)	BA17	3	3	3	2	1	1	1	
Caudate/Accumbens/Putamen	Cd/Acb/Pu	3	3	3	3				
Caudate	Cd					2	1	1	
Accumbens	Acb					1	1	1	
Putamen	Pu					2	1	1	
Globus pallidus/Putamen	GP/Pu	3	3	3	3				
Thalamus/Caudate	Th/Cd				3				
Thalamus (centromedial thalamic nucleus)	ThCeM		3	3					
Thalamus (dorsomedial thalamic nucleus)	ThMD		3	3	3				
Subthalamic nucleus	STh	3	3						
Amygdala	Amyg	3	3	3	3				
Hippocampal formation (head)	Hip head	3	3	3	3				
Hippocampal formation	Hip	3	3	3	3				
Mesencephalon	Mes	3	3	3	3				
Metencephalon rostral	Met rostr	3	3	3					
Metencephalon caudal	Met caud	3	3	3					
Myelencephalon or Myelencephalon/Medulla	Myen/Med	3	3	3	3				
Pons	Pons				2				
Cerebellum	Cereb	3	3	3	3	4	1	1	2
Spinal cord cervical	SC cerv	3	3	3					
Spinal cord thoracic	SC thor	3	3	3					
Spinal cord lumbar	SC lumb	3	3	3					
Spinal cord sacral	SC sacr	3	3	3					
Dorsal root ganglion	DRG	3							
Cerebrospinal fluid	CSF	1	1	1					
Retina	Retina					1	1	1	
Peripheral organs/tissues									
Blood	Blood				1	1	1	1	1
Heart	Heart	3	3	3	3	2	1	2	
Lung	Lung	3	3	3	3	2	2	1	
Stomach	Stomach	3	3	3	3		1	1	
Jejunum	Jejunum						1	1	
Liver	Liver	3	3	3	2	2	1	2	4
Pancreas	Pancreas	3	3	3	2	2	2	2	
Kidney	Kidney	3	3	3	3		2	2	4
Ovary	Ovary		3						
Testis	Testis	3		3	3				
Thyroid gland	Thyroid	3	3	3					
Adrenal gland	Adren							1	
Spleen	Spleen	3	3	3	3	2	2	1	
Skeletal muscle (psoas or sartorius or temporal)	Sk muscle	1		1		2	4	2	4
White adipose tissue (periumbilical subcutis)	Wh adipose					1	2	1	

For each spinal cord region, the three tissue pieces comprised either two pieces of grey matter and one piece of white matter, or *vice versa*. HD1-3 skeletal muscle= psoas; HD1-2 heart= left ventricle; HD1 right kidney; HD1 left kidney; HD1-2: pancreas= head; HD1 lung = left lower lobe; HD2 lung = right lower lobe; HD5-8 skeletal muscle = sartorius or temporal (HD5: 2x sartorius; HD6 2x sartorius and 2x temporal; HD7 1x sartorius and 1x temporal; HD8 4x sartorius); HD7 kidney = medulla (one piece) and cortex (one piece); HD7 pancreas = head (one piece) and tail (one piece); HD5 and HD6 heart = left ventricle; HD7 heart = left ventricle (one piece) and right ventricle (one piece). HD5-7 cerebellum = cerebellar vermis. HD5-7 lung= right lobe. HD6, HD7 stomach = fundus. Unless specified as above, tissue anatomical regions were not further distinguished.

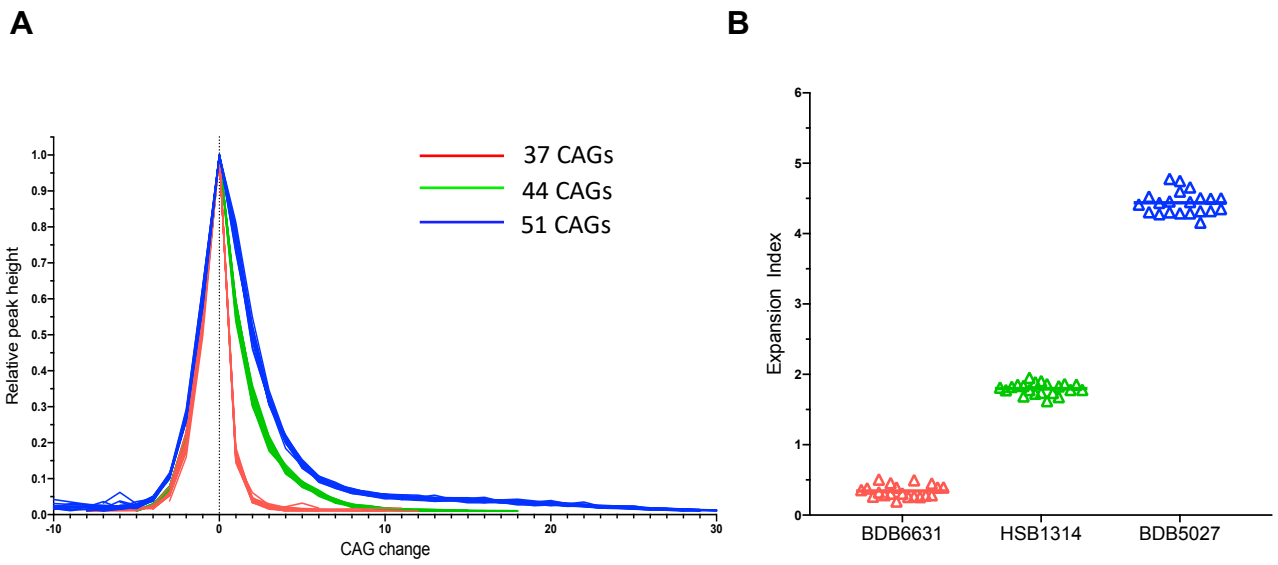
Figure S1:*HTT* CAG repeat GeneMapper profiles in CNS and peripheral tissues of individuals HD1, HD2 and HD3.



Representative GeneMapper traces are shown for a subset of tissues analyzed, representing the range of expansions observed. The red vertical line indicates the modal CAG allele of 43 or 44 for HD1 and HD2 tissue, respectively, and the modal allele of 53 for the majority of HD3 tissues.

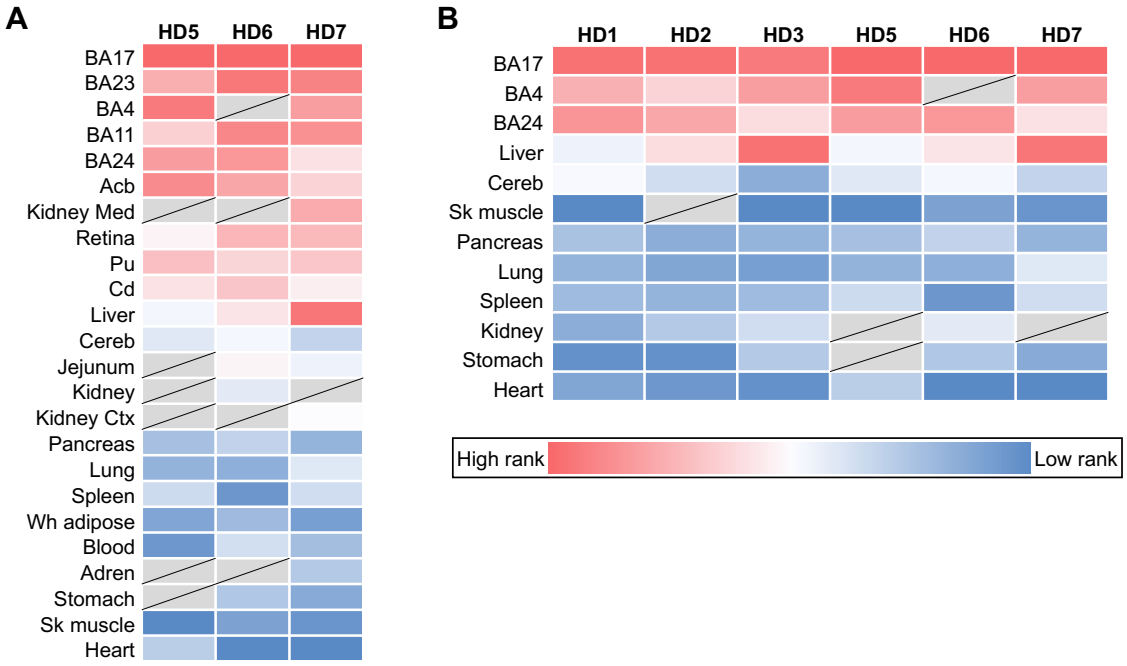
OB: olfactory bulb; BA9: prefrontal cortex; BA4: primary motor cortex; BA3.1.2: primary somatosensory cortex; Tmp: temporal pole; BA17: primary visual cortex; Cd/Acb/Pu: caudate accumbens putamen; ThMD: dorsomedial thalamic nucleus; Amyg: amygdala; Hip: hippocampal formation; Mes: mesencephalon; Cereb: cerebellum; SC cerv: spinal cord cervical; CSF: cerebrospinal fluid; Sk muscle: psoas skeletal muscle.

Figure S2: Technical variation across replicate PCRs from three cortex samples



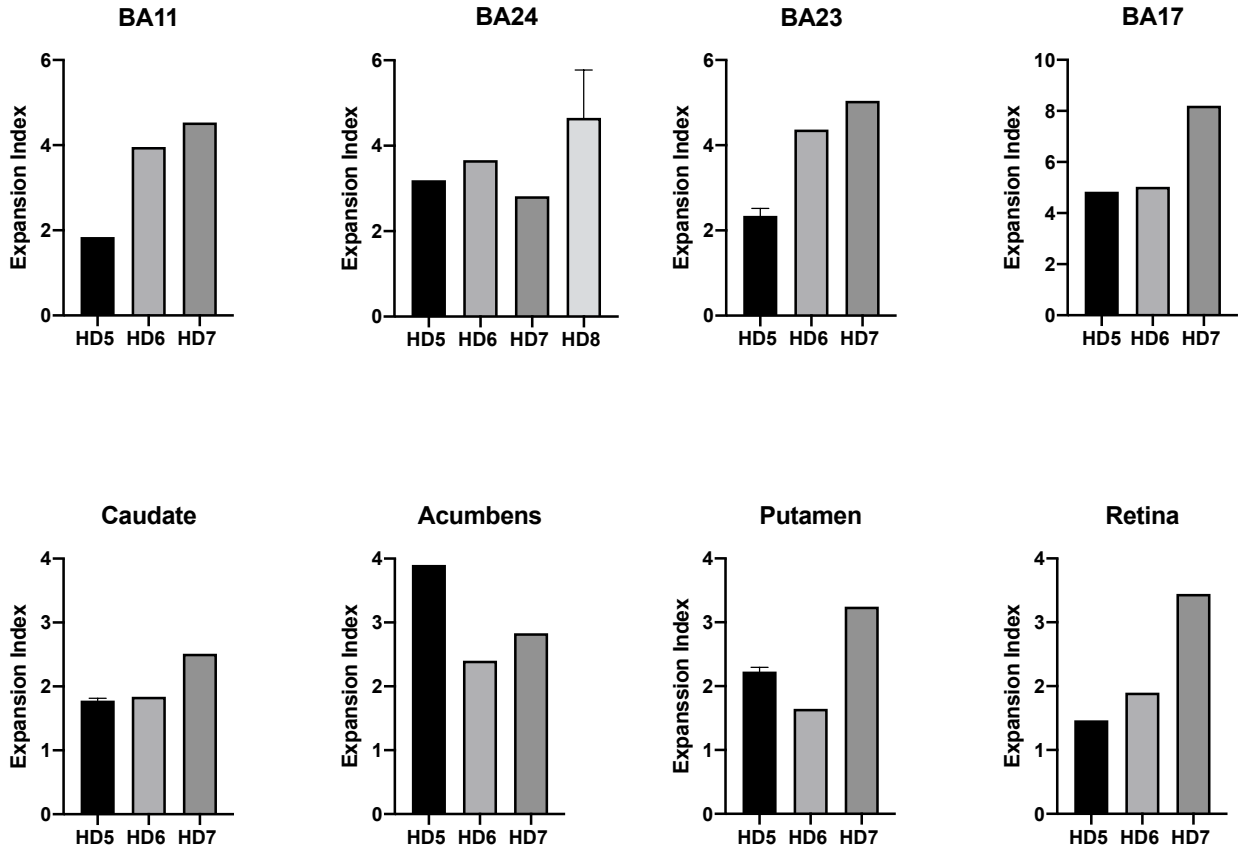
Technical variation across replicate (CAG37 N=19, CAG44 and CAG51 N=20) *HTT* CAG repeat PCR products amplified from postmortem HD cortex (BA9) genomic DNA extracted from three HD patient samples. **A**: GeneMapper peak height data, normalized to the height of the modal allele and showing repeat length change relative to the modal CAG allele. **B**: Expansion Indices (1% relative peak height threshold) calculated from the GeneMapper peak height data. Black bars show mean of the replicates.

Figure S3: Ranking of Expansion Indices in samples HD5-7 and across the two independent studies.



Mean Expansion Indices were ranked for each individual and ranks displayed as heat map. Tissues are displayed in order of mean rank. Dark red = most expanded; white = median; dark blue = least expanded; grey = tissue not available. **A**. All tissues from HD5-7 (HD8 was excluded as the tissue number was limited. **B.** All tissues shared between HD1-3 and HD5-7 with at least four individuals for each tissue (see Table S1).

Figure S4: Comparison of Expansion Indices in different brain regions across HD individuals HD5-HD8.



BA11: straight gyrus; BA24: anterior cingulate/midcingulate cortex; BA23: postcingulate cortex: BA17: primary visual cortex. For replicate measurements mean+/- S.D is plotted.

Figure S5: Point-to-point plots of Expansion Indices in all samples from all tissues relative to those in blood, liver, skeletal muscle and CSF.

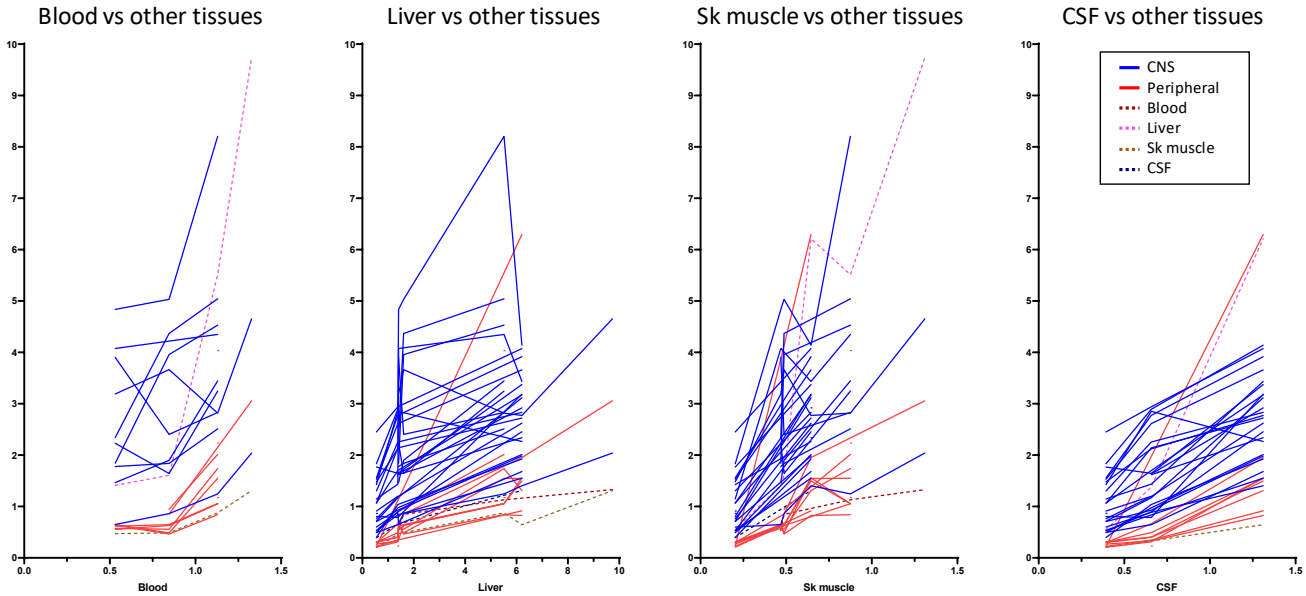
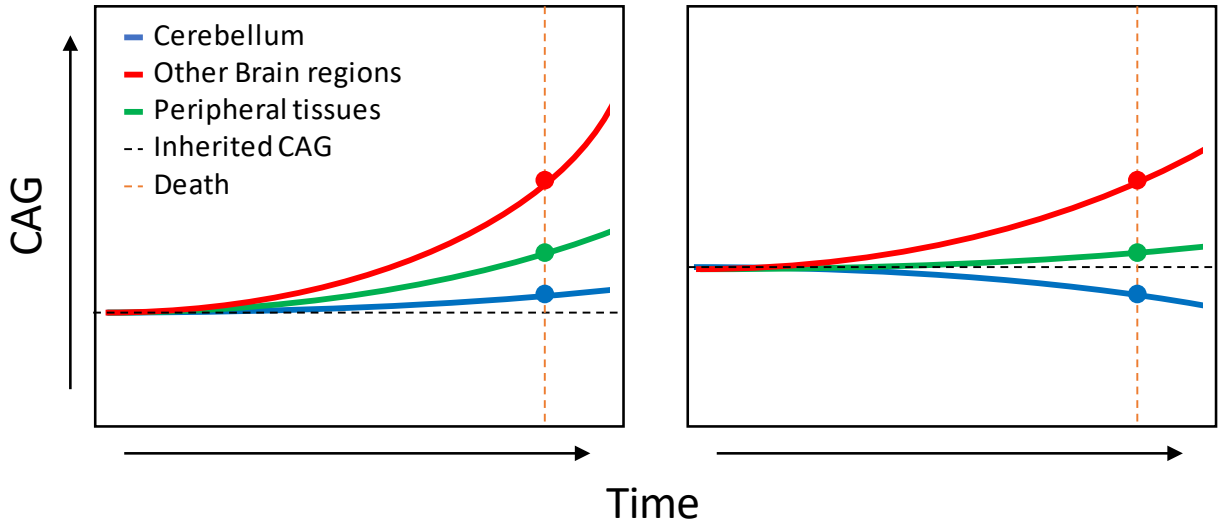


Figure S6: Graphs to illustrate possible scenarios of cerebellar repeat dynamics in the juvenile-onset individual HD4.



Left: hypothetical inherited CAG ~100 and net CAG expansion in cerebellum, at a slow rate relative to other brain regions and peripheral tissues
Right: hypothetical inherited CAG ~130, with net CAG contraction in cerebellum.