

Supplemental Table: Main findings of each arm of the study.

Arm of Study	Main Findings
Unrelated cohort (N=781)	<ul style="list-style-type: none"> • 424 (77.7%) FECD cases were CTG18.1exp+ • 18 (7.7%) controls were CTG18.1exp+ • A diagnosis of FECD associated with CTG18.1 expansion status ($p=2.48 \times 10^{-44}$) and CTG18.1_L ($p=3.38 \times 10^{-46}$) • CTG18.1exp+ status associated with FECD severity ($p=5.62 \times 10^{-7}$) • FECD severity associated with CTG18.1_L among FECD cases ($p=8.8 \times 10^{-4}$)
Family cohort (N=112 families; 331 subjects)	<ul style="list-style-type: none"> • 146 (67.0%) FECD cases were CTG18.1exp+ • 27 (23.9%) controls were CTG18.1exp+ • A diagnosis of FECD associated with CTG18.1 expansion status ($p=6.23 \times 10^{-13}$) and log₁₀ (CTG18.1_L) ($p=8.0 \times 10^{-6}$) • Penetrance of FECD associated with CTG18.1exp+ was 85.4%
CTG18.1exp+ families (N=87 families; 249 subjects)	<ul style="list-style-type: none"> • Penetrance of FECD increased by age quartile (range: 44.4-86.2%) • No sex difference in penetrance of FECD ($p=0.08$)
Parent-offspring relationships from CTG18.1exp+ families (N=62 families; 106 offspring)	<ul style="list-style-type: none"> • CTG18.1 expansion transmitted to offspring in 62 (58.5%) parent-offspring relationships • 41 (66.1%) CTG18.1exp+ offspring exhibited the FECD phenotype • No significant difference in transmission of the expanded repeat by parental sex ($p=0.62$) • 8 (18.2%) offspring who did not inherit the CTG18.1 expansion exhibited the FECD phenotype
Intergenerational instability among CTG18.1exp+ offspring (N=62)	<ul style="list-style-type: none"> • 8 (12.9%) had change in CTG18.1_L ≥ 50 repeats • 3 (4.8%) expansions of 1,900 repeats • 5 (8.1%) contractions (range: -900 to -1,900 repeats)