

Supplemental materials

Figure legends

Table 1. Patient Demographics

Table 2. Correlation coefficient (R^2) and significance (P) values for linear regression analysis comparing disease duration and age with clinical outcome measures: mFARS, ADL, T25FW, and 9HPT. The entire cohort (All pts) and sub-groups of patients with either less than or equal to 700 triplets or greater than 700 triplets were analyzed.

Figure 1. Linear regression analysis comparing GAA1 length with blood frataxin level (A) and age of onset (AoO) (B) in the entire cohort (All samples) and for subgroups stratified by GAA1 length. Correlation coefficient (R^2) values are plotted for each subgroup. Black data points represent results for patients with less than or equal to the GAA1 cutoffs listed on the X-axis e.g. less than or equal to 200 triplets, 300 triplets...Red data points represent results for patients with greater than the GAA1 cutoffs listed on the X-axis e.g. greater than 200 triplets, 300 triplets...Filled data points= $p < 0.01$; open data points=n.s. Linear regression plots comparing GAA1 length with blood frataxin level (C) and AoO (D) for the entire cohort stratified by GAA1 less than or equal to 700 triplets (gray circles, black fit line) or more than 700 triplets (white circles, red fit line).

Figure 2. Linear regression analysis comparing disease duration with clinical outcome measures: modified Friedreich ataxia rating scale (mFARS) (A), activities of daily living (ADL) (B), timed 25-foot walk (T25FW) (C), and 9-hole peg test (9HPT) (D). Light gray data points and black fit line=results for the entire cohort (all pts). Dark gray data points and red fit line=results for patients with greater than 700 triplets.

Figure 3. Prevalence of cardiomyopathy (A), scoliosis (B), and diabetes (C) for the entire cohort (n=all) and for sub-cohorts stratified by GAA1 length.

Supplemental figures

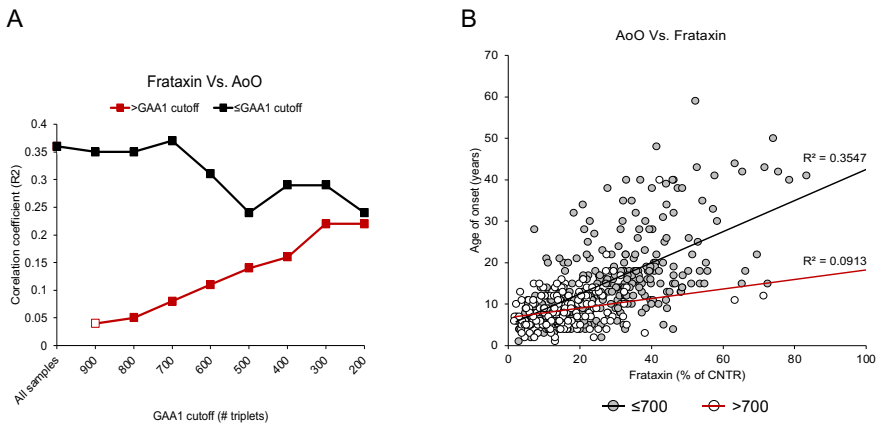


Figure S1. (A) Linear regression analysis comparing blood frataxin level with age of onset in the entire cohort (All samples) and for subgroups stratified by GAA1 length. Correlation coefficient (R²) values are plotted for each subgroup. Black data points represent results for patients with less than or equal to the GAA1 cutoffs listed on the X-axis e.g. less than or equal to 200 triplets, 300 triplets...Red data points represent results for patients with greater than the GAA1 cutoffs listed on the X-axis e.g. greater than 200 triplets, 300 triplets...Filled data points=p<0.01; open data points=n.s. (B) Linear regression plot comparing blood frataxin level with age of onset for the entire cohort stratified by GAA1 less than or equal to 700 triplets (gray circles, black fit line) or more than 700 triplets (white circles, red fit line).

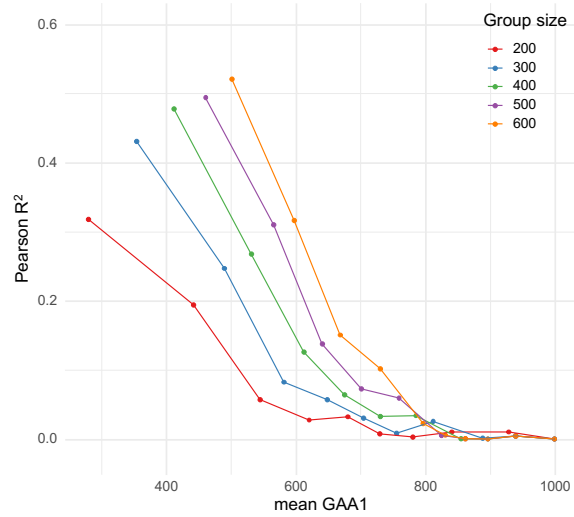


Figure S2. Rolling correlation analysis of GAA1 vs age of onset. After sorting patients by increasing GAA1, the correlation coefficient was calculated for patients 1-200, 100-300, etc. (red line), and analogously for 5 different groups sizes (200-600). Pearson's R2 was then plotted over mean GAA1 repeat length within the respective groups.

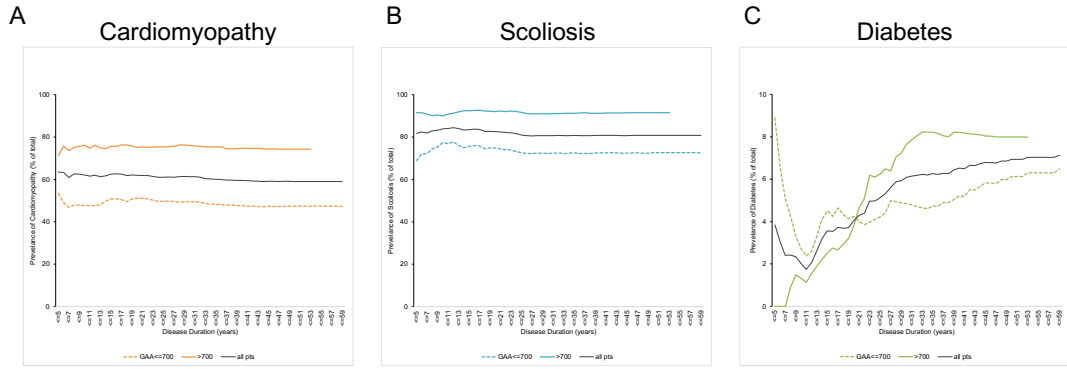


Figure S3. Prevalence of cardiomyopathy (A), scoliosis (B), and diabetes (C) for the entire cohort (all pts/black lines) and for sub-cohorts of patients with ≤ 700 triplets (dashed lines) or greater than 700 triplets (solid colored lines) plotted based on disease progression.