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GeneDx Validation for Repeat Expansion Disorders Assay

Dear Sir or Madam,

GeneDx, Inc., a CAP/CLIA-accredited genetic testing laboratory, frequently develops new testing for genes and disorders for which there are limited testing options available. Part of the assay development process relies on the evaluation of DNA samples from individuals with a known genetic disorder and/or specific type of genetic change to validate the effectiveness of the assay. GeneDx is currently developing assays for a subset of repeat expansion disorders and is looking for samples from patients with a known diagnosis and positive genetic test results for any one of the following genes/disorders:

Disorder	Gene
Friedreich's ataxia	*FXN**
DRPLA	*ATN1*
SCA10	*ATXN10*
SCA12	*PPP2R2B*
SCA17	*TBP*
SCA31	*BEAN*
SCA36	*NOP56*
SCA37	*DAB1*

*We're seeking participants with specific repeat lengths between 66 and 400 repeats.

If you choose to participate in these studies you will be financially compensated $100 for a blood sample and $25 for a buccal (cheek swab) sample. Blood samples are preferred and a home blood draw can be arranged at a time that is convenient for you. We request that you provide a copy of the original lab report confirming your diagnosis, but we can work with your physician to acquire these documents. All required paperwork for participation in the study and compensation for your time will be discussed at enrollment.

Thank you very much for your time and for any assistance you might provide.

Sincerely,

Ben Solomon, MD
Managing Director, GeneDx