Clinicians Guide: Genetic Testing for Hereditary Ataxia

Fulgent offers a variety of genetic testing panels to target more than 160 genes associated with hereditary ataxia, including cerebellar ataxias, episodic ataxias, autosomal recessive ataxias, autosomal dominant ataxias and X-linked ataxias. Available panels include repeat expansion analysis and sequencing analysis, including deletion/duplication. Our genetic counselors are available to help you select the most appropriate test for your patient.

Who is this test for?

Patients with a known or suspected family history of ataxia with presentation of symptoms consistent with hereditary ataxia. Signs of ataxia include incoordination of gait with poor coordination of hands, speech, and/or eye movement.

What are the potential benefits for my patient?

Identifying the specific genetic cause of your patient's ataxia can help confirm a clinical diagnosis and/or determine medical management for your patient. It can also provide information about clinical course of disease and illuminate potential risk for close relatives of the patient.

Which panel should I order?

We provide several methods of testing for hereditary ataxia. When the genetic etiology of the disease is completely unknown, repeat expansion analysis is often performed first, as it is the most common mechanism for hereditary ataxia. Repeat expansion analysis may be ordered per gene, or as a 14-gene panel. Fulgent also offers sequencing analysis of additional genes commonly associated with ataxia. Potential options for ordering are included below, but the best method for each patient will depend on their specific family and medical history.

1 Ordering Repeat Expansion Analysis (either by selecting genes, or ordering the full 14-gene panel) → Diagnostic? → Yes → No further testing necessary

Diagnostic? → No → **2 Ordering Panel (one of the following):**
a. Early-onset Ataxia Panel
b. Late-onset Ataxia Panel
c. Ataxia Panel (combines early and late-onset panels)

Fulgent also offers the ability to customize panels based on your patient's needs.

For example, if repeat expansion analysis has been completed for some, but not all genes, you may choose to order the sequencing Ataxia Panel plus a select number of repeat expansion genes. Details on number and type of genes in each panel can be found on the reverse side of this flyer.

4978 Santa Anita Ave, Suite 205, Temple City, CA 91780 | www.FulgentGenetics.com | info@fulgentgenetics.com | **P** +1 626.350.0537 | **F** +1 626.454.1667

D# FLY-Clinicians Guide: Hereditary Ataxia-2022

Ataxia Repeat Expansion Analysis: 14 genes

Gene	Abbreviation	Condition
FXN	FXN	Friedreich's Ataxia
ATXN1	SCA1	Spinocerebellar Ataxia 1
ATXN2	SCA2	Spinocerebellar Ataxia 2
ATXN3	SCA3	Spinocerebellar Ataxia 3
CACNA1A	SCA6	Spinocerebellar Ataxia 6
ATXN7	SCA7	Spinocerebellar Ataxia 7
ATN1	DRPLA	Dentatorubral-pallidoluysian atrophy
TBP	SCA17	Spinocerebellar Ataxia 17
NOP56	SCA36	Spinocerebellar Ataxia 36
ATXN10	SCA10	Spinocerebellar Ataxia 10
ATXN8OS	SCA8	Spinocerebellar Ataxia 8
BEAN1	SCA31	Spinocerebellar Ataxia 31
PPP2R2B	SCA21	Spinocerebellar Ataxia 12
FMR1	FXTAS	Fragile X-associated tremor/ataxia syndrome

Coverage

96% at 20x
for sequencing

Turnaround Time

3-5 weeks for repeat expansion analysis

3-5 weeks for sequencing analysis

Sample Requirements

(kits available upon request)

Acceptable samples

- Blood, two 4mL EDTA tubes, lavender top (preferred)
- Extracted DNA, 3µg in TE buffer
- Buccal swab or saliva

Ataxia Sequencing Panels

Early-onset Ataxia panel: 134 genes

Appropriate for patients with symptom onset over the age of 18 years. This panel does not include repeat expansion analysis.

ABCB7, ABHD12, ACO2, AFG3L2, AHI1, ALDH5A1, ALS2, ANO10, APTX, ARL13B, ATCAY, ATL1, ATM, ATP8A2, B9D1, BBS1, BBS12, BSCL2, C12orf65, C19orf12, CA8, CAMTA1, CC2D2A, CEP290, CEP41, CLCN2, CLN5, CLPP, COQ2, COQ6, COQ8A, COQ9, COX20, CPLANE1, CSTB, CWF19L1, CYP27A1, CYP7B1, DNAJC19, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, FA2H, FBXL4, FGF14, FLVCR1, FXN, GALC, GBA2, GFAP, GJC2, GOSR2, GRID2, GRM1, GSS, HEPACAM, INPP5E, ITPR1, KCNA1, KCNC3, KCND3, KCNJ10, KIF1A, KIF1C, KIF5A, KIF7, LAMA1, MLC1, MRE11, MTPAP, NDUFS1, NDUFS7, OFD1, OPA1, OPA3, OPHN1, PAX6, PDSS1, PDSS2, PEX10, PEX7, PHYH, PLP1, PNKD, PNKP, PNPLA6, POLG, POLR3A, POLR3B, PRKCG, PRRT2, RORA, RPGRIP1L, RRM2B, RUBCN, SACS, SETX, SIL1, SLC16A2, SLC1A3, SLC2A1, SLC52A2, SLC52A3, SLC9A6, SNX14, SPAST, SPG11, SPR, SPTBN2, STUB1, TCTN1, TCTN2, TCTN3, TDP1, TGM6, TMEM216, TMEM231, TMEM237, TMEM240, TMEM67, TPP1, TTPA, TUBB4A, TWNK, TYMP, VAMP1, VLDLR, WDR73, WDR81, WFS1, WWOX, ZNF423

Late-onset Ataxia panel: 57 genes

Appropriate for patients with symptom onset over the age of 18 years. This panel does not include repeat expansion analysis.

ABHD12, AFG3L2, ALS2, ANO10, APTX, ATL1, C19orf12, CACNA1A, CACNB4, CCDC88C, CLCN2, CYP27A1, CYP7B1, DNMT1, EEF2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL4, ELOVL5, FGF14, FMR1, FXN, GFAP, ITM2B, ITPR1, KCNC3, KCND3, KIF5A, NOL3, NOTCH3, PDYN, PEX7, PHYH, POLG, PRKCG, RORA, RRM2B, SETX, SLC1A3, SPAST, SPG11, SPG7, STUB1, SYNE1, SYT14, TGM6, TMEM240, TTBK2, TTPA, TUBB4A, TWNK, TYMP, VAMP1, WASHC5

Ataxia panel: 153 genes

This panel includes all genes within the early- and late-onset panels. This panel does not include repeat expansion analysis.

ABCB7, ABHD12, ACO2, AFG3L2, AHI1, ALDH5A1, ALS2, ANO10, APTX, ARL13B, ATCAY, ATL1, ATM, ATP8A2, B9D1, BBS1, BBS12, BSCL2, C12orf65, C19orf12, CA8, CAMTA1, CC2D2A, CEP290, CEP41, CLCN2, CLN5, CLPP, COQ2, COQ6, COQ8A, COQ9, COX20, CPLANE1, CSTB, CWF19L1, CYP27A1, CYP7B1, DNAJC19, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, FA2H, FBXL4, FGF14, FLVCR1, FXN, GALC, GBA2, GFAP, GJC2, GOSR2, GRID2, GRM1, GSS, HEPACAM, INPP5E, ITPR1, KCNA1, KCNC3, KCND3, KCNJ10, KIF1A, KIF1C, KIF5A, KIF7, LAMA1, MARS2, MLC1, MRE11, MTPAP, NDUFS1, NDUFS7, OFD1, OPA1, OPA3, OPHN1, PAX6, PDSS1, PDSS2, PEX10, PEX7, PHYH, PLP1, PNKD, PNKP, PNPLA6, POLG, POLR3A, POLR3B, PRKCG, PRRT2, RORA, RPGRIP1L, RRM2B, RUBCN, SACS, SETX, SIL1, SLC16A2, SLC1A3, SLC2A1, SLC52A2, SLC52A3, SLC9A6, SNX14, SPAST, SPG11, SPR, SPTBN2, STUB1, TCTN1, TCTN2, TCTN3, TDP1, TGM6, TMEM216, TMEM231, TMEM237, TMEM240, TMEM67, TPP1, TTPA, TUBB4A, TWNK, TYMP, VAMP1, VLDLR, WDR73, WDR81, WFS1, WWOX, ZNF423