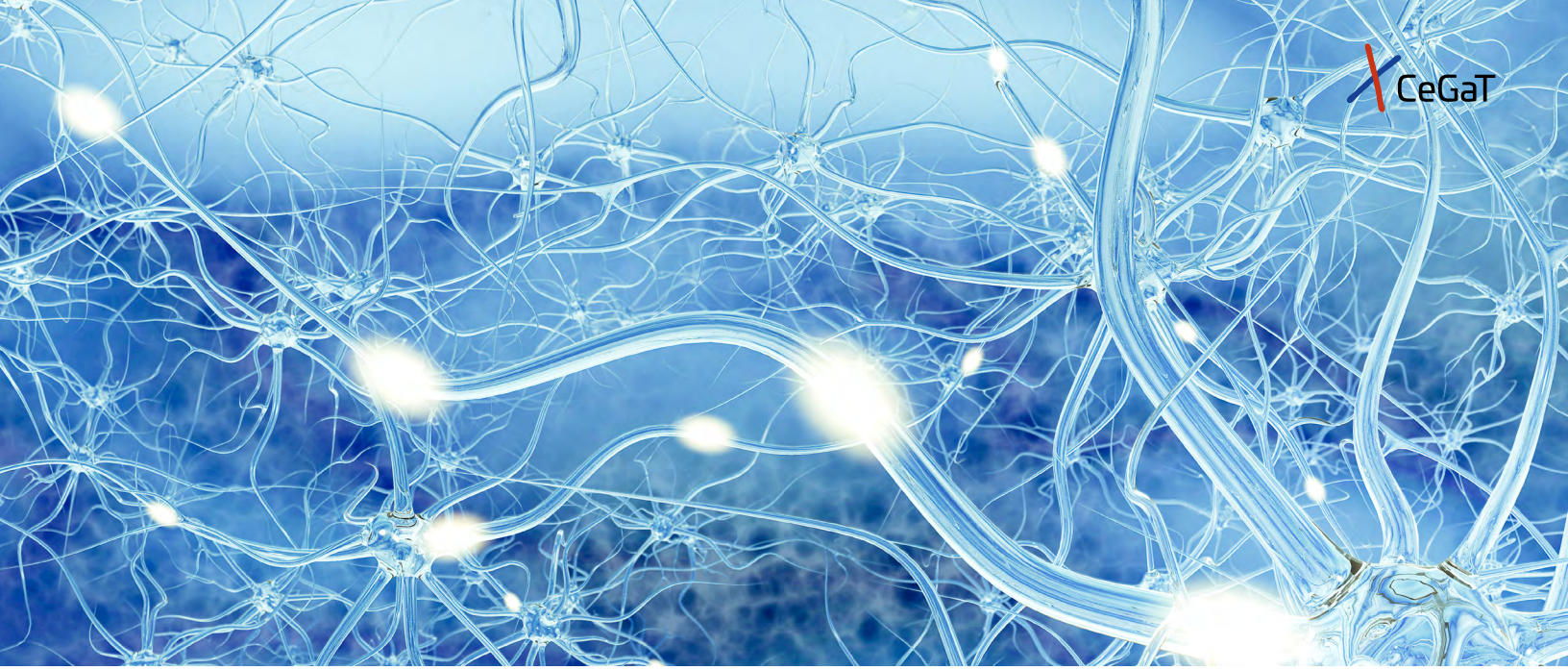

Genetic Diagnostic for Ataxia

At CeGaT, we are dedicated to advancing the diagnosis of Ataxia through cutting-edge genetic expertise and technology. Our comprehensive and accessible testing services support patients, physicians, and healthcare partners in every step of the diagnostic journey. By offering affordable and reliable solutions, we help accelerate accurate diagnoses and open the door to personalized treatment options.

Benefit from Our Genetic Diagnostics for your Ataxia Patients!

High Coverage of Genes

Broad panel including 110 genes and detection of SNVs, Indels and CNVs in coding and noncoding clinically relevant regions as well as repeat expansion detection for selected genes, including *FGF14* (SCA27B)

Over 15 Years of Experience in Panel Diagnostics

- CAP/CLIA accredited laboratory, every step of the process performed in-house.
- Panels are regularly updated with current knowledge
- Fast TAT, medically urgent cases prioritized without additional cost
- Easy ordering process with myCeGaT portal, with diagnostic and customer support at any step
- VUS Re-Evaluation included

Customizable Panel Options

Create a panel that fits best to your individual patients needs from our expert curated gene sets

Broad Insurance Coverage - Low Maximum Out-of-Pocket Costs

Flat rate payment for Medicare, Medicaid, Blue Cross Blue Shield or TriCare patients

ExomeXtra®: Superior In-House Designed Enrichment

- All protein-coding regions of the genome
- Clinically relevant RNA transcripts
- >46,000 intergenic and intronic positions associated with genetic disease according to ClinVar or HGDM
- Pharmacogenetically relevant variants in selected genes
- Higher coverage of the mitochondrial genome
- Genome-wide backbone for the detection of CNVs (replaces array CGH)
- Sequencing on NovaSeq™ X Plus at 100-150x

CeGaT Corporation | 2430 N-Halsted #110 | Chicago, IL 60614 | phone: 312.783.5225 | www.cegat.com

The Broadest Genetic Insights for Ataxia Patients

Our Panel

Ataxia and Differentialdiagnosis (NDD14, 110 Genes)

ABCB7, ABHD12, AFG3L2, ANO10, APTX, ATCAY, ATG7, ATM, ATP1A3, ATP8A2, CA8, CACNA1A, CACNA1G, CACNB4, CAMTA1, CAPN1, LCN2, CLN6, COA7, COQ8A, CP, CTBP1, CWF19L1, CYP27A1, DAB1, DARS2, DNAJC5, DNMT1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL4, ELOVL5, EPM2A, EXOSC5, FAT2, FGF14, FLVCR1, FXN, GDAP2, GFAP, GOSR2, GRID2, GRM1, HEXA, HEXB, ITPR1, KCNA1, KCNC3, KCND3, KCNJ10, KCNN2, KIF1C, MARS2, MRE11, HLRC1, NKX6-2, NOP56, NPC1, NPC2, PDYN, PIK3R5, PITRM1, PLA2G6, PMPCA, PNKP, PNPLA6, POLG, POLR3A, POU4F1, PPP2R2B, PRICKLE1, PRKCG, PRRT2, PUM1, RNF170, RNF216, RUBCN, SACS, SCN2A, SCYL1, SETX, SIL1, SLC1A3, SLC2A1, SLC52A2, SLC52A3, SLC9A1, SNX14, SPG7, SPTBN2, STUB1, SYNE1, TDP2, GM6, THG1L, TMEM240, TPP1, TTBK2, TTPA, TWNK, VAMP1, VLDLR, VPS13D, VPS41, WDR81, WWOX, XRCC1

SCA1, SCA2, SCA3, SCA6, SCA7, SCA17 repeat analyses and of FXN repeat analysis is part of our standard procedure.

Selected Publications:

- Spinocerebellar Ataxia Type 35 Caused by a New TGM6 Variant: Video Documentation of a German Family, Maass *et. al.*, 2023
- Heterozygous UCHL1 loss-of-function variants cause a neurodegenerative disorder with spasticity, ataxia, neuropathy, and optic atrophys, Park *et al.*, 2022
- New Nonsense Variant c.2983G>T; p.Glu995* in the CACNA1A Gene Causes Progressive Autosomal Dominant Ataxia. Saathoff *et. al.*, 2021
- Clinical variability in ataxia-telangiectasia. Lohmann *et. al.*, 2015
- Novel ATM mutation in a German patient presenting as generalized dystonia without classical signs of ataxia-telangiectasia. Kuhm *et. al.*, 2015
- Two novel mutations of the SETX gene and ataxia with oculomotor apraxia type 2., Lechner *et al.*, 2015
- Mutations in the sodium channel gene SCN2A cause neonatal epilepsy with late-onset episodic ataxia., Schwarz *et. al.*, 2016

Contact Us for Your Genetic Testing Service:

Eric R. Miller

Head of Business Development & Sales – North America

Email eric.miller@cegat.com
Mobile 312.287.7545

www.cegat.com/us

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