National Ataxia Foundation

Genetic Testing & Counseling: Ataxia Practice Recommendations for Clinicians

Based on *Practice Recommendations for Genetic Testing of Ataxias* (Srinivasan & Mook et al., 2025), this handout contains evidence-based, consensus-driven recommendations to support clinicians in evaluating patients with Ataxia in order to provide timely evaluation and genetic diagnosis for patients and at-risk relatives.

Purpose of Document: Genetic testing is becoming a vital diagnostic tool for ataxia. To support clinicians, these practice recommendations were developed to offer a practical framework for timely and effective genetic evaluation. Pages 3-4 contain a proposed workflow for evaluation of hereditary ataxia and key points to include in genetic counseling, meant as a quick reference guide.

What is Ataxia?

While the term “ataxia” is used to describe symptoms of incoordination that may occur in a variety of disease processes, the term “Ataxia” may also used to label a heterogenous group of neurodegenerative diseases. The degenerative ataxias (referred to herein as Ataxia) result from hereditary or sporadic genetic variants that ultimately lead to degeneration of the cerebellum, cerebellar pathways, and/or the spinal cord.

Why Should Genetic Testing Be Considered?

For Patient:

- Diagnosis
- Influence on treatment and/or avoidance of contraindicated treatment
- Avoidance of additional unnecessary testing & procedures
- Surveillance of comorbidities
- Prognosis
- Eligibility for clinical trials or other research
- Psychological
  - Closure
  - An answer to WHY
  - Access to support organizations/community

For Patient’s Family Members:

- Accurate risk assessment
- Access to predictive genetic counseling and/or testing if desired
  - Reproductive decision making
  - Career and life planning
  - Lifestyle changes
  - Anticipation of medical needs

PO Box 27986, Golden Valley, MN 55427 www.ataxia.org naf@ataxia.org 763.553.0020

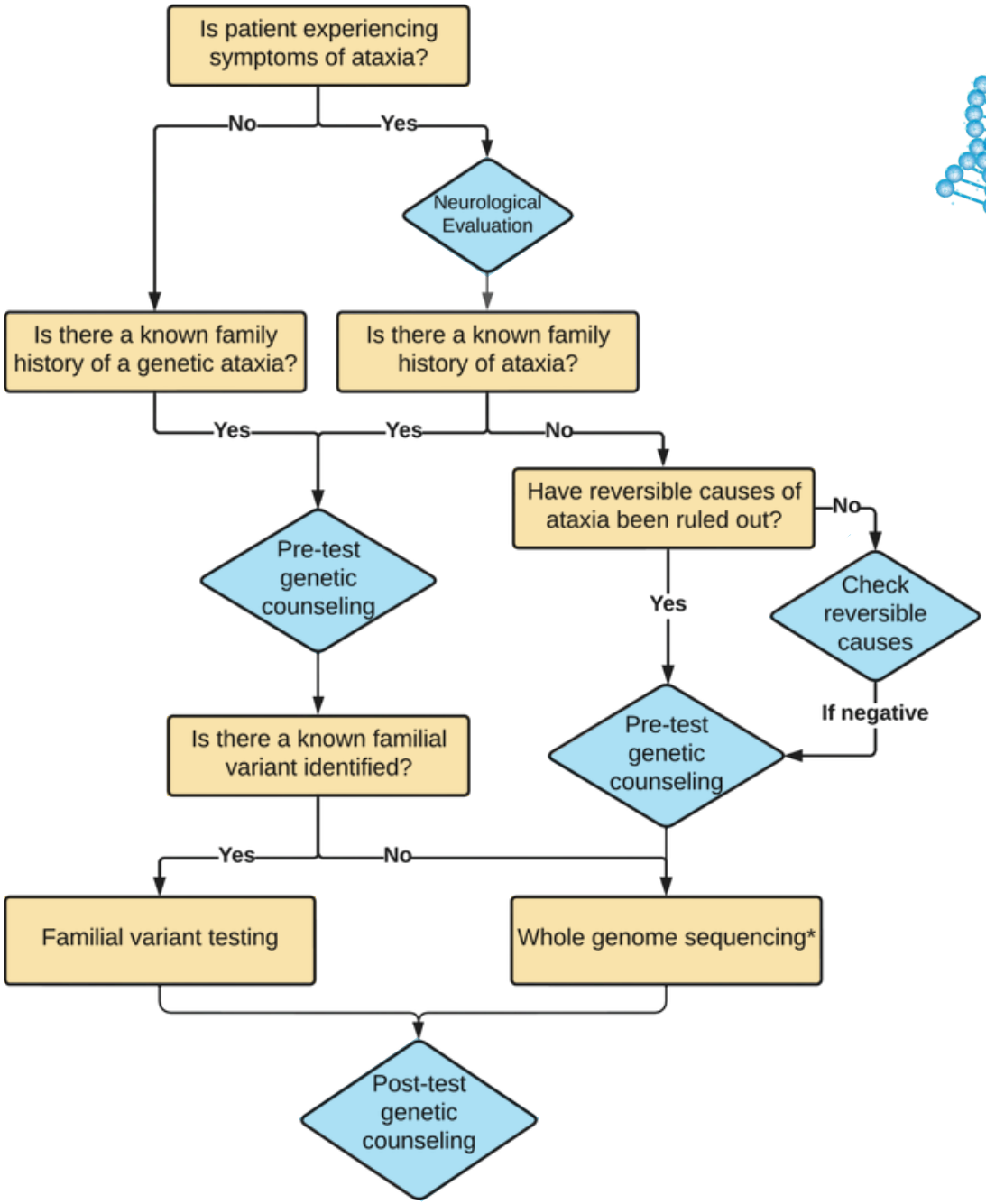
Is a Patient Ready to Consider Genetic Testing?

It is important to assess patient readiness for genetic testing. The psychological status of every patient should be considered. This may include asking about a history of depression, anxiety, and suicidal ideation, and inquiry made as to whether they are seeing a therapist and their sources of support. While not a formal requirement, a referral should be made at the discretion of the ordering provider to a therapist/psychiatrist as indicated and potentially deferring testing until appropriate mental health care is established.

When Should Genetic Testing Be Considered?

Diagnostic	Performed for an ataxic individual to either establish or confirm a suspected diagnosis. Testing may be requested for a variety of reasons, including the need to determine an explanation for symptoms, a desire to definitively name one's disease, and inform healthcare and life decisions.
Predictive	Performed for an asymptomatic individual with a family history of ataxia. This may be requested for a variety of reasons, including future planning for marriage, reproduction, career, home, insurance,finances, travel or simply a need to resolve uncertainty. Although there are currently no direct medical benefits from predictive testing, there are lifestyle modifications, such as cardiovascular exercise, which is shown in both humans and animal models to ameliorate symptoms and potentially slow progression.
Reproductive	Performed when an individual with a known genetic ataxia or at risk of a genetic ataxia seeks genetic testing either prior to (preconception) or during a pregnancy (prenatal). This may be requested to make decisions about testing a pregnancy or reproductive options, such as IVF or donor gametes. If a pregnancy is tested, results may be used to decide whether or not to continue a pregnancy. Therefore, persons with a known personal or family history of a genetic ataxia who wish to test a pregnancy should be referred to a genetic counselor prior to or early in a pregnancy.
Pediatric	Performed when a child (individual under age 18) is symptomatic for an ataxia syndrome to either establish or confirm a suspected diagnosis. Testing may be requested to determine an explanation for symptoms and inform healthcare decisions.

Ataxia Evaluation Workflow



Genetic Testing Strategy

We recommend that initial genetic testing should include whole genome sequencing (WGS) with the ability to accurately size short tandem repeats to reduce patient burden, account for newly discovered genes, and maximize currently available technology.

If WGS as a first approach is fiscally impossible or otherwise not feasible, then a dedicated repeat expansion panel with eventual reflex to WGS would be appropriate.

Ordering genetic testing should be expedited if there is high clinical suspicion of a genetic ataxia with an FDA-approved treatment.

Genetic testing of minors should only be offered when the minor is determined to be symptomatic by a pediatric neurologist with expertise in ataxia. Testing of asymptomatic minors may be considered if there is an approved treatment in childhood and there is a positive family history with an identified or suspected pathogenic variant(s).

Genetic Counseling Considerations

Pre-Test Counseling

- ☐ Collect medical history, including patient-perceived symptoms and time course of ataxia
- ☐ Collect a three-generation family history and ages of onset in affected individuals
- ☐ Provide personal risk assessment based on inheritance pattern(s) of suspected ataxia/neurological condition(s).
- ☐ Test selection
  - Determine if ordering proband only versus duo, trio, or quad testing with additional family members
  - If relevant, review additional information that the patient may opt in or out of receiving (e.g. secondary or incidental findings)
- ☐ Review types of tests and results
  - Repeat expansion analysis: positive, negative, intermediate
  - Sequencing and deletion/duplication analysis: positive, negative, VUS
- ☐ Discuss benefits, risks, and limitations of genetic testing, and personal utility
  - Review cost of genetic testing, billing options and whether clinic or patient will need to contact lab/insurer to determine if testing will be fully, partially, or not at all covered
  - Review possible implications of genetic test results for insurance coverage (e.g. life)
- ☐ Timing of genetic testing
  - Discuss if timing is optimal if requested close to holidays or major lifecycle events
- ☐ For patients who do not undergo testing, more general resources on ataxia should be provided
- ☐ Obtain informed consent, if proceeding with testing
- ☐ Determine plan for communication of results with consideration of modality and timing with recognition of potential immediate release of results to patients due to the 2021 CURES Act

Post-Test Counseling

- ☐ Patients should be provided with a copy of their results report and appropriate counseling as dictated by their results type

- ☐ For a negative or uncertain result, patients should be encouraged to periodically reach out for updates and additional testing recommendations (if any) due to the evolving knowledge of genetic ataxias.

- ☐ For a positive result:
  - Discuss any changes to management, including availability of FDA-approved therapies or treatment, research opportunities including clinical trials
  - Discuss implications for relatives and recommendations for genetic testing

Provide disease-specific resources relevant to the patient including support groups. If no specific ataxia is identified, more general resources can be provided.

References

1. Srinivasan, S. R., Mook, A. D., Rochman, M., Chen, J. Y. H., Mu, W., Wilmot, G. R., Rosenthal, L. S., & Uhlmann, W. R. (2025). Practice recommendations for Genetic Testing of Ataxias. Annals of Clinical and Translational Neurology, 12(12), 2398–2409.

Notably, the authors of these practice recommendations practice in the United States. These guidelines are U.S.-specific, though may be relevant internationally depending on local healthcare systems and laws. Additionally, novel methodological approaches and improvements in understanding of the genetic basis of ataxias may impact implementation of these recommendations.

Handout developed by Amy Mook, MS, MPH, CGC and Michelle Rochman, MS, CGC in collaboration with the National Ataxia Foundation. Other resources for clinicians can be found at ataxia.org/provider-resources

Last updated April 2026