

Pre-Test (ASD)

# 1. Genetic Testing Checklist

Supports physicians in discussing genetic testing and ensuring informed decision-making



## DIAGNOSTIC CRITERIA

*Patient has met criteria for ASD genetic testing*

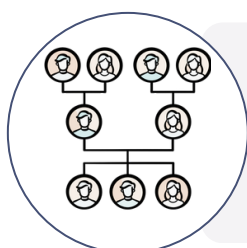
Clinical diagnosis of autism spectrum disorder or global developmental delay, intellectual disability, or speech delay AND Incomplete, outdated or absence of genetic testing



## GENETIC TESTING

*Understanding Genetic Testing for ASD*

- ASD is diagnosed clinically.** A mixture of environment and genetic factors underly the condition. Over 100 genes, however, are known to play a role. Genetic testing can identify an underlying cause in 40% of individuals. Knowing genetic changes that cause a particular case of ASD could connect patient with organizations and families with the same mutation and guide personal medical management or future planning.
- Standard of Care:** Multiple medical professional societies including the Canadian Paediatric Society (CPS), American Academy of Pediatrics (AAP), American College of Medical Genetics (ACMG) and American Academy of Clinical Psychiatrists (AACCP) have recommended genetic testing, specifically CMA as the standard of care.
- Genetic testing is always a choice.** Genetic counseling remains available should you want to discuss this in more detail.



## FAMILY HISTORY

Complete three generation pedigree: ([How To Draw Your Family History Pedigree Video](#))



## UNDERSTANDING BASIC GENETICS

- Share educational video
  - [English](#)
  - [Spanish](#)
- Provide pamphlet
  - Key Points: Genetic testing is a test that looks in your cells to identify an underlying genetic component to ASD
  - Genes make us who we are.
  - Tests: [Chromosomal Microarray](#) & [ASD/ID Panel](#)
  - Genetic results may be **positive (pathogenic)**, **negative (benign)** or **uncertain (VUS- variant of uncertain significance)**.
  - VUS result may or may not be associated with phenotype. Indicates evidence is insufficient or conflicting and not associated with disease. Classification of VUS might change over time.
  - Genetic testing is protected by the Genetic Non-Discrimination Act (GNDA 2020)