PRETEST ASD
Genetic Testing Checklist

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of autism spectrum disorder or global developmental delay, intellectual disability, or speech delay AND Incomplete, outdated or absence of genetic testing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

1. **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.
2. **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 (AACP) have recommended genetic testing, specifically Fragile X, CMA as the standard of care.
3. 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

1. **Share educational video**
   a. **English**
   b. **Spanish**
2. **Provide pamphlet**
3. **Key Points:**
   a. Genetic testing is a blood test to help identify an underlying genetic component to ASD
   b. Genes make us who we are.
   c. Tests: **Chromosomal Microarray** and **Fragile X**
   d. Genetic results may be positive (pathogenic), negative (benign) or uncertain (VUS- variant of uncertain significance).
   e. Genetic testing is protected by the Genetic Non-Discrimination Act (GNDA 2020)

ORDERING TESTING

1. Ordering Via **Connect Care**
2. Ordering Guide: **Chromosomal Microarray (CMA): Information for Ordering**
3. Requisition form for Reference
   a. **Cytogenetics Requisition form (CMA):**
   b. **Molecular Genetics Requisition Form (Fragile X testing, or FMR1 disorders)**

TIMELINE

Genetic test results are usually available in 6-8 weeks after the blood sample is received in the lab

RESULTS DISCLOSURE

1. **NEGATIVE:** Please discuss results with the patient and assure they received a copy of their results as well as an appropriate handout. Results will live in EHR.
2. **POSITIVE/VUS:** Patients with a pathogenic results or a variant of uncertain significance (VUS) should be referred for an appointment at the Genomic Psychiatry Consultation to ensure that results are reported under the guidance of a genetic professional and management is reviewed. Results will live in EHR. Referrals to the Genomic Psychiatry consult service using ***this form.***