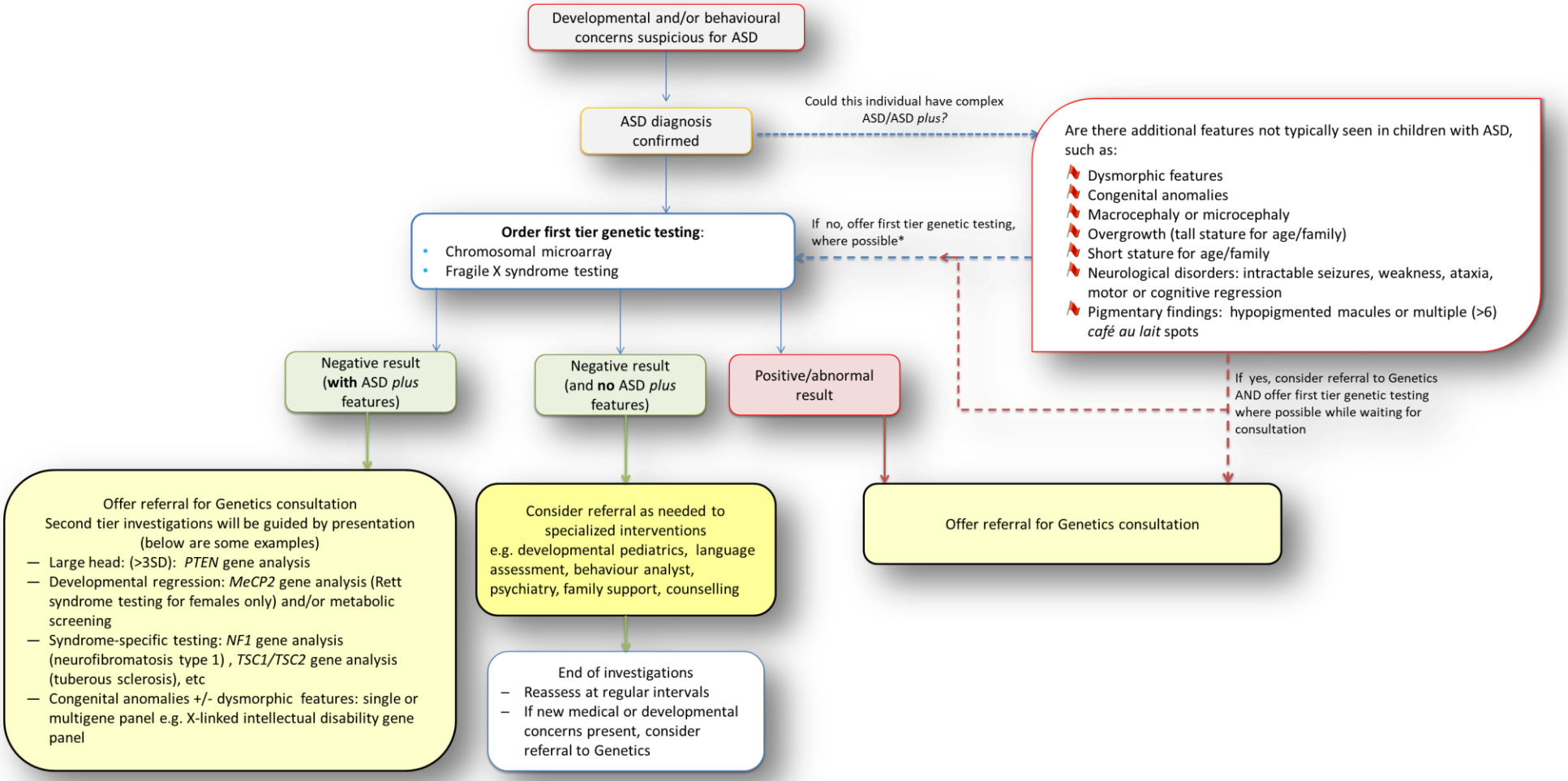
GENETICS OF AUTISM SPECTRUM DISORDER

A road map of possible genetic tests and consultations for the individual with autism spectrum disorder (ASD).



Autism spectrum disorder (ASD) is a complex, genetically influenced disorder, affecting about 1 in 68 children. ASD is highly variable both in presentation and in etiology. In families where one child has ASD, the risk to subsequent siblings is about 10-19%. About 15-40% of individuals with ASD will have an identifiable contributing genetic cause.

Once an ASD diagnosis is confirmed, a provider can consider the red flags above to determine if the individual has isolated ASD or ASD *plus*. A genetics referral could be made at that time. Alternatively, a provider who is comfortable providing pre-test counselling and post-test follow-up could begin the first tier genetic testing and refer upon receipt of results.

First tier genetic investigations for all individuals with ASD are chromosomal microarray and fragile X syndrome testing. If an individual is determined to have ASD *plus* (signifying additional co-morbidities such as congenital anomalies, dysmorphic features, neurological symptoms (e.g. seizures)) additional investigations may be considered. A genetic diagnosis can potentially lead to guideline-based surveillance and management, tailored treatment options, opportunities to participate in clinical trials, information regarding natural history and prognosis, familial testing and accurate recurrence risk counselling. Primary care providers who feel confident providing pre- and post –test counselling may be able to arrange first tier genetic investigations prior to or concurrent with referral for genetic consultation.

*Local laboratories may limit which providers can order first tier investigations. Contact your local genetics centre for more information and advice.

For community support and research opportunities for your patient and his/her family, see Autism Speaks Canada.

For a concise, evidence-based resource on ASD and genetics including benefits and limitations of genetic testing for ASD, please see the GEC-KO on the run. For a more comprehensive review and complete reference list please see the GEC-KO Messenger.

For an excellent review on ASD diagnosis in the primary care setting, please see Anagnoustou and colleagues.