# Genetic Assessment: Frequently Asked Questions

**Etiologic assessment is often helpful in planning preventive care, treatment, and management strategies. Many adults whose developmental disabilities (DD) are of unknown origin may benefit from etiologic assessment or reassessment.**

Contact a local Genetics Centre for help in deciding whether to refer, and for the referral criteria and protocol.

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| **How do I find the nearest Genetic Centre for my patient?** | Contact information for Genetic Centres in Canada is available at **https://cagc-accg.ca/**. |
| **Are there ways of determining the likelihood of a patient's having a genetic etiology for his/her DD, so as to prioritize whom I should consider referring?** | The chances of individuals having a genetic etiology for their DD generally range from greater to lesser likelihood in the following order:<br>• Family history of DD.<br>• Congenital malformations.<br>• Dual diagnosis (DD and co-occurring mental illness). If possible, patients should be seen by a knowledgeable psychiatrist and/or clinical geneticist who can identify those more likely to have a genetic etiology, including those with a specific pattern of behaviours or with a specific psychiatric diagnosis.<br>• Severe to profound DD, congenital malformation(s).<br>• Mild to moderate DD, congenital malformation(s). |
| **Why might a genetic assessment be helpful?** | **Optimal medical management**<br>• A tailored medical and psychosocial management approach to address physical and mental health issues can be developed once the etiology is established. For example, people with Down syndrome have an increased probability of developing thyroid disease throughout their lifespan and will benefit from earlier and more regular screening than guidelines for the general population recommend.<br>• Identifying a genetic etiology can have health management consequences for other family members. For example, in the fragile X syndrome, pre-mutation carrier males and females have the potential to develop fragile X-associated tremour/ataxia syndrome and females have an increased risk of premature ovarian failure.<br><br>**Family reasons**<br>• The patient and other family members may want information about the cause of the DD and the risk of recurrence within the family.<br>• There can be substantial guilt about having a child with a DD. Knowing the cause can relieve parental guilt and provide reassurance. As well, with this knowledge, family members will be aware of and be able to address issues related to specific syndromes, such as by connecting with syndrome-specific organizations. |

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| **Is there a special referral form? What information is needed on the referral?** | Contact or check the website of your nearest genetic centre.<br>• Helpful information to include in a referral:<br>– Detailed reason(s) for referral.<br>– Any previous genetic test results and the date of the test(s).<br>– Copies of other pertinent investigations (e.g., MRI, echocardiogram).<br>– Name and contact information of the Substitute Decision Maker, if needed, to provide consent for genetic testing. |
| **Are there some tests I should do, as a family physician, prior to sending a patient with DD to a genetic centre?** | • Currently some genetic centres request that fragile X molecular testing and karyotype (chromosome analysis) be done before the patient is seen for genetic assessment. Microarray analysis is a much more sensitive test that is replacing the karyotype. Call the genetics centre closest to you, or visit its website, for further information as to which tests are appropriate to do prior to referral, and for access to requisitions.<br>• The reasons for these tests and for a genetics referral should be discussed with the individual, his/her family, and/or the Substitute Decision Maker in order to obtain appropriate consent for undertaking them.<br>• Salient clinical information should be included on the requisition form.<br>• If you order a test and the results are abnormal (including any type of variant found on microarray or chromosome analysis and pre-mutation carrier status for fragile X), consult a clinical geneticist regarding implications and for patient and family feedback and counseling. Referral to a genetics centre is highly recommended. In some cases, parental studies may be undertaken through the genetics centre to assist in interpretation of results. |
| **Is genetic testing covered by health insurance?** | • In Canada, genetic assessment and some medical tests are covered by the provincial health care plan. The clinical geneticist may request some tests that are performed only outside Canada and may seek prior approval for payment for them from the provincial authorities.<br>• Private labs may charge a fee to take and transport a blood sample for some tests (e.g., molecular testing). Check with the service providers in your area. |
| **Which diagnoses can be detected through genetic testing?** | • Over 900 diagnoses are currently possible through genetic testing, a number that increases annually.<br>• Genetic testing diagnoses include single gene mutations, whole or partial chromosome duplications and deletions (including micro-duplications and micro-deletions), imprinting defects, and mitochondrial disorders.<br>• Relevant to the population with DD, examples of diagnoses from genetic testing include Down syndrome, fragile X syndrome, Prader-Willi syndrome, Williams syndrome, Smith-Magenis syndrome and 22q11.2 deletion syndrome. Some syndrome-specific reviews are available at **www.ncbi.nlm.nih.gov/sites/genetests**. |

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| **Which diagnoses are not primarily genetically determined?** | • DD can be caused by factors other than genetic conditions, including infections, exposure to toxins/teratogens, and perinatal hypoxemia or trauma. Cerebral palsy and fetal alcohol spectrum disorders are examples.<br>• Such a diagnosis does not preclude the person from also having a genetic syndrome. |
| **What happens when the patient and family members go for genetic testing and counseling?** | • The patient and family members will meet with a genetic counselor and/or geneticist who will obtain a detailed patient and family medical history, as well as explain the reason for the genetic assessment.<br>• A physical exam is usually done by the geneticist. This may include measurement of salient physical features (e.g., facial ones), and photography of such features (with appropriate consent) for the individual's medical record.<br>• Possible genetic diagnoses may be reviewed and appropriate tests to help determine a diagnosis will be discussed. Consent to retrieve records may be requested.<br>• Information will be given so that the patient and family can provide informed consent with respect to the proposed genetic testing.<br>• Lab tests will usually be done, including blood or urine tests. In some circumstances it may be helpful to obtain other investigations such as a skin punch biopsy, X-rays, ultrasound, CT or MRI scans. Referral to other specialists may also be recommended. |
| **How are test results communicated?** | • Results are normally reported to the referring physician and communicated to the patient, family and/or Substitute Decision Maker by the geneticist. This may include providing a genetic diagnosis and offering further counseling.<br>• Genetic changes causing DD can occur sporadically or may be hereditary. Potential consequences of a hereditary disorder, the likelihood of developing it or transmitting it to one's children, and whether there are means to prevent it or lessen any of these effects would be discussed.<br>• Reproductive options should also be discussed at the request of the patient and/or family members. |
| **How does management of the patient proceed?** | • Management may be through the geneticist, family physician or other specialist, depending on the condition, the needs of the patient, and available resources.<br>• If no etiology is determined, periodic reassessment is recommended. |

## Resources

**www.geneticresourcesontario.ca** This online guide for health care providers and the public includes a list of genetic centres and specific genetic services in Ontario.

**www.mountsinai.on.ca/care/family-medicine-genetics-program/family-medicine-genetics-program** This Canadian website gives primary health care providers current practical information regarding screening and prevention of hereditary disorders.

**www.ncbi.nlm.nih.gov/sites/genetests** Provides "Gene Reviews," expert-authored peer-reviewed disease descriptions.

**www.askthegen.org** This website of the Department of Human Genetics at Emory University, "Ask the Geneticist," answers questions about genetic concepts, and the etiology, treatment, research, testing and predisposition to genetic disorders.

**"Genetics through a Primary Care Lens" at www.genetests.org/servlet/access?id=8888892&key=VVeQo6NaqTUT8&fcn=y&fw=W5jm&filename=/tools/index.html** US National Institutes of Health Genetic Tools website provides background information, teaching cases, and links to other resources for primary care providers, including a section on Developmental Delay and Genetics at a Glance.

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