Genetic Assessment of Adults with Intellectual and Developmental Disabilities

Frequently Asked Questions

Introduction

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

This information sheet answers common questions that family physicians may have about including genetic testing professionals involved in their patients' care. It provides information for finding the nearest Canadian genetic centre, indications for referral and how to make a referral, and outlines information to give the person with an intellectual and developmental disability and their support persons regarding the referral process.

Supporting materials

i. **Genetics Education Canada – Knowledge Organization.**
Not-for-profit genetics and genomics education for non-genetics health professionals.
https://geneticseducation.ca/

ii. **Canadian Association of Genetic Counsellors.**
List of medical genetic centres located across Canada. https://www.cagc-accg.ca/

iii. **The Family Medicine Genetics Program. Primary care resources.**
Mount Sinai Hospital, Toronto, Ontario
http://www.mountsinai.on.ca/care/family-medicine-genetics-program/family-medicine-genetics-program

iv. **Gene Reviews**
University of Washington, Seattle
www.genereviews.org

v. **Orphanet**
Provides information on rare diseases and orphan drugs. www.orpha.net

These supporting materials are hosted by external organizations and the accessibility of these links cannot be guaranteed. The DDPCP will make every effort to keep these links up to date.

Genetic Assessment of Adults with Intellectual and Developmental Disabilities: Frequently Asked Questions

How do I find the nearest genetic centre for my patient?

- Contact a local genetic centre for help in deciding whether to refer, and for the referral criteria and protocol. Contact information for genetic centres in Canada is available at the Canadian Association of Genetic Counsellors website: https://cagc-accg.ca/

What is the likelihood of a patient having a genetic etiology for his/her IDD, to prioritize whom I should consider referring?

The chances of having a genetic etiology for IDD are generally increased if there is:

- Severe to profound IDD, congenital malformation(s), epilepsy, or complex medical presentation.
- Mild to moderate IDD, congenital malformation(s), epilepsy, or complex medical presentation.
- Family history of IDD and/or consanguinity.
- Dual diagnosis (IDD 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.

Why might a genetic assessment be helpful?

Optimal medical management

- It may be possible to develop a tailored medical and psychosocial management approach to address physical and mental health issues 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.
- Identifying a genetic etiology can have health management consequences for family members. For example, in the fragile X syndrome, pre-mutation carrier males and females have the potential to develop fragile X-associated tremor/ataxia syndrome and females have an increased risk of premature ovarian insufficiency.

Family reasons

- The patient and other family members may want information about the cause of the IDD and the risk of recurrence within the family.
- There can be substantial guilt about having a child with a IDD. Knowing the cause can relieve parental guilt and provide reassurance. As well, with this knowledge, family members may be able to find support by connecting with syndrome-specific organizations.

Is there a special referral form? What information is needed on the referral?

Contact or check the website of your nearest genetic centre at the Canadian Association of Genetic Counsellors website: https://cagc-accg.ca/

- Information to include in a referral:
  - Detailed reason(s) for the referral
  - Any previous genetic test results and the date of the test(s)
  - Copies of other pertinent investigations (e.g., MRI, echocardiogram)
  - Name and contact information of the Substitute Decision Maker, if needed, to provide consent for genetic testing

Are there tests I should do, as a family physician, prior to sending a patient with IDD to a genetic centre?

- Currently some genetic centres request that fragile X molecular testing and microarray be done before the patient is seen for genetic assessment. Microarray analysis is a much more sensitive test that has replaced the karyotype (chromosome analysis). Contact the genetics centre closest to you, for further information as to which tests are appropriate to do prior to referral, and for access to requisitions.
- 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.
- Salient clinical information should be included on the requisition form.
- If you order a test and the results are abnormal (including any type of variant found on microarray 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 out of province or outside Canada and may seek prior approval for payment for them from the provincial authorities.
- 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 2500 genes have been associated with syndromic and non-syndromic IDD, including autism spectrum disorder. This number is continuing to increase as our knowledge expands.
- Genetic tests can identify single gene pathogenic variants (mutations), whole or partial chromosome duplications and deletions (including microduplications and microdeletions), imprinting defects, and mitochondrial disorders.
- Relevant to the population with IDD, 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 GeneReviews, University Washington: www.genereviews.org

Which diagnoses are not primarily genetically determined?

- IDD can be caused by factors other than genetic conditions, including infections, exposure to toxins/teratogens, and perinatal hypoxemia or trauma. Some forms of cerebral palsy and fetal alcohol spectrum disorders are examples.
- 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.
- 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.
- Possible genet*ic diagnoses may be reviewed* and appropriate tests to help determine a diagnosis will be discussed. Potential complexity in interpreting test results may be reviewed (such as uncertain or unexpected results). Consent to retrieve records may be requested.
- Information will be given so that the patient and family can provide *informed consent* with respect to the proposed genetic testing.
- *Lab tests* will usually be done, including blood and/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. ▸ Genetic changes causing IDD 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. ▸ 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. ▸ If no etiology is determined, periodic reassessment may be recommended.

References

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