Exome Sequencing in Cardiogenetics

Genetic testing for cardiac-related indications can be offered by physician specialists (e.g., geneticists, cardiologists, cardiac surgeons) who diagnose, manage, and/or treat individuals with known or suspected inherited cardiomyopathies, arrhythmias, or other cardiological conditions with a genetic basis.

When cardiogenetic diagnostic certainty is high, especially with supporting ancillary test results, the recommended testing approach is to select the most appropriate single or multi-gene panel (see Genetics Guidance for additional details). However, exome sequencing (ES) is the recommended first-tier test for the following individuals:

Funded Cardiac-related Indications for ES

Individuals who meet at least 2 of the following criteria:

- Congenital structural heart defect
- Arrhythmogenic condition and/or cardiomyopathy, not believed to be secondary to a structural heart defect
- Aortopathy, not believed to be secondary to a structural heart defect
- Other clinical features suggestive of a genetic syndrome (e.g., major non-cardiac congenital anomaly, dysmorphic features, global developmental delay/intellectual disability)

If a patient is eligible for ES as outlined above, **chromosomal microarray analysis (CMA) should be ordered concurrently with ES if not previously completed.**

Additional Support

For further assistance in test selection, test ordering, result interpretation, and management recommendations, please consider the following options as needed:

- Access lab requisitions and additional details at Genome-wide Sequencing Ontario
- Send a referral to your local genetics clinic (Genetics Clinics in Ontario)
- Submit an eConsult request to connect with a genetics specialist through OTNhub