



WHAT TO CONSIDER WHEN REFERRING FOR GENETIC TESTING FOR HATTR?



HF Update 2021 – Corridor Consult

Nathalie Bolduc, MSc, CCGC, CGC
Certified Genetic Counsellor, Genolife

Nowell Fine MD SM FRCPC
Cardiologist, University of Calgary



LEARNING OBJECTIVES

What should you consider before referring a patient for genetic testing?

How should you manage the findings and what are the implications for patients and their families?

DISCLOSURES

Nathalie Bolduc

- Director of Business Development at Genolife, a private genetic services company
- Consultant for the Prevention and Genetics Program at the Quebec Breast Cancer Foundation
- Speaker Fees: Akcea/Ionis, Dialogue

Nowell Fine

- Consulting Fees/Honoraria: Pfizer, Akcea/Ionis, Alnylam, Takeda, Sanofi-Genzyme, Astra-Zeneca, Bayer
- Clinical Trials: Pfizer, Eidos, Akcea/Ionis, Alnylam
- Speaker Fees: Pfizer, Akcea/Ionis, Alnylam
- Research Grants: Pfizer, Akcea/Ionis, Alnylam, Eidos

GENETIC TEST - POSSIBLE BENEFITS

Establish a diagnosis

- Understand the cause of medical / family history
- *Consider who is the best person to test

Appropriate management

- Enables access to targeted management
- Positive reinforcement of behaviors / lifestyle habits
- Allows life/reproductive planning decisions
- Identify at-risk family members
- Identify non-carriers



POSSIBLE DISADVANTAGES

Results may indicate a probability or carry uncertainty

- Incomplete penetrance
- Variability in symptoms presentation
- Variants of uncertain significance
- « Recent » gene with few guidelines

Difficult psychological reactions (anxiety, guilt)

Impact on family (difficulties, conflicts, familial distress)



Canada passes genetic ‘anti-discrimination’ law

May 4 2017 |

Canada’s House of Commons has passed a new law that prevents corporations from demanding genetic information from potential employees or customers.

The law, known as ‘Bill S-201’, makes it illegal for companies to deny someone a job if they refuse a genetic test, and also prevents insurance companies from making new customer policies conditional on the supply of genetic information. Insurance companies will no longer be able to solicit genetic tests so as to determine customer premiums.

NEWS

Supreme Court of Canada rules genetic non-discrimination law is constitutional

By [Leslie MacKinnon](#). Published on Jul 10, 2020 11:51am



GENETIC TESTING FOR ATTR

Canadian Journal of Cardiology 36 (2020) 322–334

Society Position Statement

Canadian Cardiovascular Society/Canadian Heart Failure Society Joint Position Statement on the Evaluation and Management of Patients With Cardiac Amyloidosis

RECOMMENDATION

7. For patients with a diagnosis of ATTR cardiac amyloidosis, we recommend the performance of genetic testing to differentiate hATTR from wtATTR (Strong Recommendation, Moderate-Quality Evidence).

Rationale for Genetic Testing

- Prognostic
- Non-cardiac manifestations
- Family members risk
- Therapy selection




OUR APPROACH

Amyloidosis Program of Calgary



- At first appointment after ATTR diagnosis
 - Hereditary vs Wild-type subtypes explained
 - Family / support / caregivers encouraged to attend
- Genetic testing recommended
- Option of Genetic Counselling referral before testing offered
- Industry sponsored TTR gene mutation testing programs utilized
- If positive, Genetic Counselling recommended (if not yet performed)
 - Offer to discuss with family members, refer for testing if desired





WHAT TO CONSIDER WHEN REFERRING FOR GENETIC TESTING FOR HATTR?

Thank you!

Discussion / questions / comments

