Caring for rare hearts: Inherited cardiovascular disease pearls

Friday, November 8th, 2024

June C Carroll MD CCFP FCFP
Shawna Morrison MS CGC
Kirsten Bartels MSc CCGC

VANCOUVER CONVENTION CENTRE

FMF
Family Medicine Forum
Forum en médecine familiale

THE COLLEGE OF FAMILY PHYSICIANS OF CANADA
LE COLLÈGE DES MÉDECINS DE FAMILLE DU CANADA

Land acknowledgment

We acknowledge that we are presenting to you today from the shared, unceded, ancestral territories of the xʷməθkʷəy̓əm (Musqueam), Sḵwx̱wú7mesh (Squamish), and səlilwətaɬ (Tsleil-Waututh) Nations.

The Musqueam, Squamish and Tsleil-Waututh have never left their territories and will always retain their jurisdiction and relationships with the territory.

Silent Genomes Project

Reducing health care disparities and improving diagnostic success
for children with genetic diseases from Indigenous populations

"…Indigenous people often have little or no access to these [genomic] technologies, increasing the health disparity gap. Silent Genomes is a game-changing effort to address this inequity, by bringing life-changing genomic diagnosis to children while ensuring Indigenous-led governance over biological samples and health data."

Laura Arbour, *MD, MSc, MSc, FRCPC, FCCMG (Project Lead)*
Nadine R. Caron, *MD, MPH, FRCSC (Co-Lead)*
Wyeth W. Wasserman, *PhD (Co-Lead)*

Presenter Disclosure

Presenter: June C Carroll

Relationships with financial sponsors:

- Other: none

Presenter Disclosure

Presenter: Shawna Morrison

Relationships with financial sponsors:

- Other: Employee of Children's Hospital of Eastern Ontario which funds the genetics education program GECKO

Presenter Disclosure

Presenter: Kirsten Bartels

Relationships with financial sponsors:

- Other: Employee of Provincial Health Services Authority (BC)

Disclosure of Financial Support

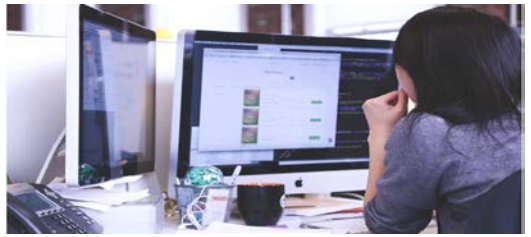
Potential for conflict(s) of interest:
Dr. Carroll and Ms. Morrison have received funding from the Children's Hospital of Eastern Ontario for travel.

Kirsten Bartels has no external support

Objectives

- Determine who to offer genetic testing and/or specialist assessment for hypertrophic cardiomyopathy (HCM), familial hypercholesterolemia (FH), heritable thoracic aortic disease (HTAD).

- Discuss clinical utility of genetic testing for, HCM, FH HTAD



- Identify where to find credible resources for genomics relevant to your practice

Cardiac Diseases with a Heritable Component

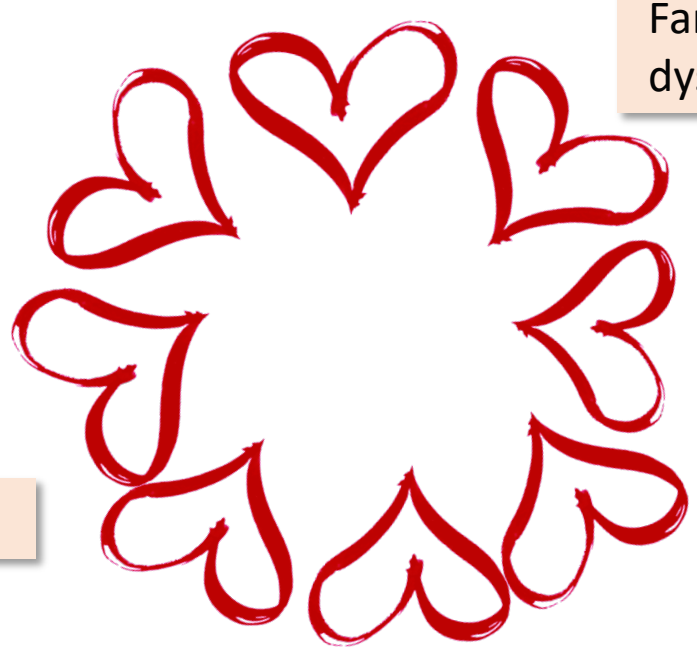
Familial Aneurysms (Thoracic)

Coronary Artery Disease

- Familial hypercholesterolemia and dyslipidemias

Congenital Heart Disease

- Isolated or syndromic



Muscular Dystrophy (MD) and Cardiomyopathy

- Duchenne and Becker MD
- Carriers of Duchenne and Becker MD
- Myotonic Dystrophy

Cardiomyopathies

- Hypertrophic Cardiomyopathy
- Dilated Cardiomyopathy
- Cardiac Amyloidosis
- Arrhythmogenic Right Ventricular Dysplasia (ARVC)

Valvular Disease

Thrombophilia

- Factor V Leiden deficiency
- von Willebrand disease

Arrhythmia Syndromes

- Atrial Fibrillation
- Long QT Syndrome

Common cardiac presentations and family histories that suggest heritable cardiac conditions:

Common cardiac presentations and family histories that suggest heritable cardiac conditions:

- Sudden death < 40 years, including SIDS

- Inherited arrhythmia or cardiomyopathy

Common cardiac presentations and family histories that suggest heritable cardiac conditions:

- Sudden death < 40 years, including SIDS
- Syncope or pre-syncope with exercise, intense emotional, stressful, or startling events

Inherited arrhythmia e.g. catecholaminergic polymorphic ventricular tachycardia (CPVT)

Common cardiac presentations and family histories that suggest heritable cardiac conditions:

- Sudden death < 40 years, including SIDS
- Syncope or pre-syncope with exercise, intense emotional, stressful, or startling events
- Seizures/Drowning/Unexplained single MVA

Long QT syndrome or another channelopathy e.g. Brugada syndrome

Common cardiac presentations and family histories that suggest heritable cardiac conditions:

- Sudden death < 40 years, including SIDS
- Syncope or pre-syncope with exercise, intense emotional, stressful, or startling events
- Seizures/Drowning/Unexplained single MVA
- Cardiac Device (ICD, pacemaker) at young age

Cardiomyopathy

Common cardiac presentations and family histories that suggest heritable cardiac conditions:

- Sudden death < 40 years, including SIDS
- Syncope or pre-syncope with exercise, intense emotional, stressful or startling events
- Seizures/Drowning/Unexplained single MVA
- Cardiac Device (ICD, pacemaker) at young age
- Cardiac disease *without* usual risk factors

Familial hypercholesterolemia

Cases

Case 1

You are seeing a 10yo boy, Adam, for a complaint of possible growing pains

His mother attends the appointment with him

She reports a new family history of hypertrophic cardiomyopathy (HCM) in Adam's paternal grandfather, and she is highly anxious about the implications for Adam especially with hockey try outs starting

Hypertrophic cardiomyopathy

Common: ~1 in 500 in the general population

Onset: variable, from infancy to late adulthood

Presentation:

- Symptoms can include shortness of breath on exertion, arrhythmias, heart failure, stroke, sudden cardiac death, often in young athletes
- Many individuals have no symptoms
- Can vary between relatives and those with the same genetic variant
- Can be isolated or rarely part of a genetic syndrome (e.g. Noonan syndrome, Fabry disease, metabolic disorders)

Sudden cardiac death (SCD):

- May be first manifestation
- HCM accounts for 13-50% of all SCDs
- Overall incidence among individuals with HCM is low (0.5-1%)

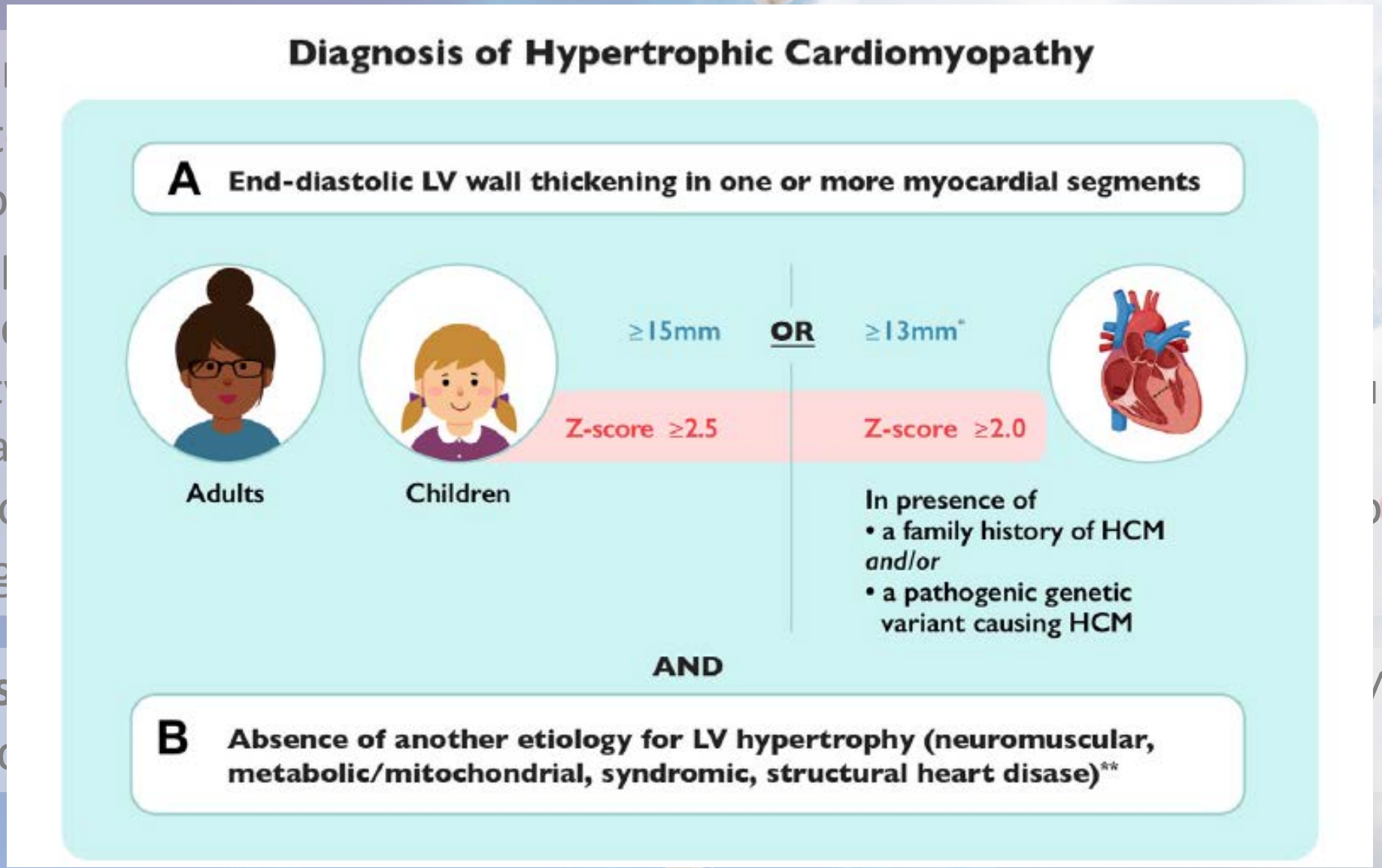
Hypertrophic cardiomyopathy

Evaluation:

- Prompted by:
  - positive family history of HCM
  - presence of symptoms such as a cardiac event
  - physical findings
    - heart murmur – typically systolic
    - pronounced apical point of maximal impulse
    - abnormal carotid pulse
    - presence of a fourth heart sound
  - abnormal findings on an echocardiogram conducted for other reasons
  - irregularities on a 12-lead ECG e.g. repolarization abnormalities, arrythmias

Diagnosis: Echocardiography, Cardiac MRI should be considered in all patients with *suspected* HCM and is complimentary to echocardiography

Hypertrophic cardiomyopathy



Canadian Cardiovascular Society, 2024

Hypertrophic cardiomyopathy

35-60% of HCM cases will have an identifiable genetic cause

Most of the responsible genes code for sarcomere and sarcomere-related proteins

Autosomal dominant inheritance is most common (50% risk to first-degree relatives)

Who should be offered genetic testing?

All patients with a confirmed or suspected clinical diagnosis of HCM.

Patients with a known pathogenic/likely pathogenic HCM-genetic variant in their family.

Deceased individuals with a suspected heritable cardiac condition as part of an autopsy.

Tip

Genetic testing must start with an affected individual.

In patients with hypertrophic cardiomyopathy, which of the following is most likely to be true regarding the genetic testing of family members?

a) Genetic testing is not recommended in asymptomatic family members
b) Only first-degree relatives should be genetically tested
c) Genetic testing can identify carriers of the disease who are asymptomatic
d) Genetic testing is useful only if both parents are affected

In patients with hypertrophic cardiomyopathy, which of the following is most likely to be true regarding the genetic testing of family members?

a) Genetic testing is not recommended in asymptomatic family members
b) Only first-degree relatives should be genetically tested
c) Genetic testing can identify carriers of the disease who are asymptomatic
d) Genetic testing is useful only if both parents are affected

Why offer genetic testing for HCM?

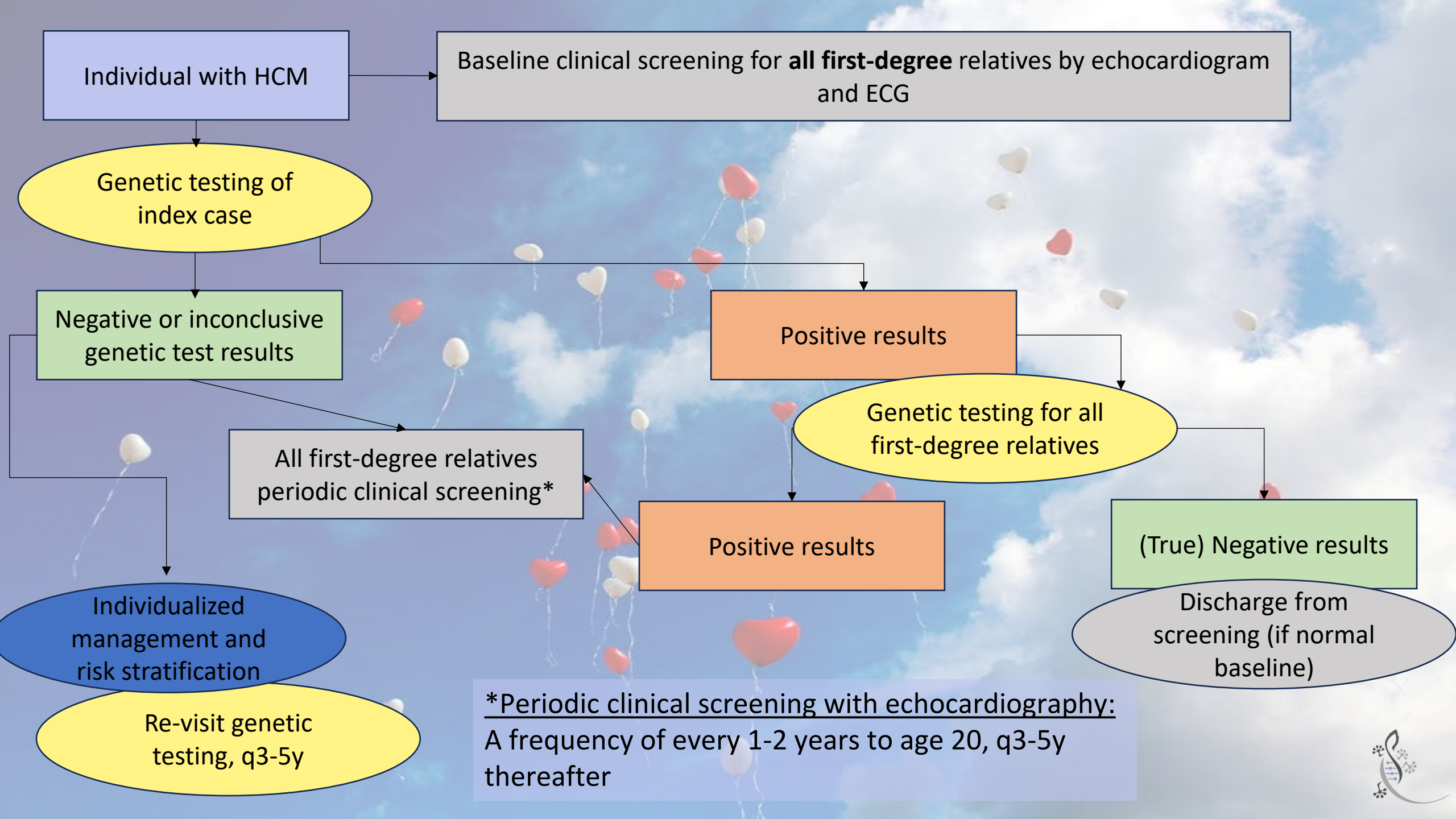
Genetic testing for HCM can help with:

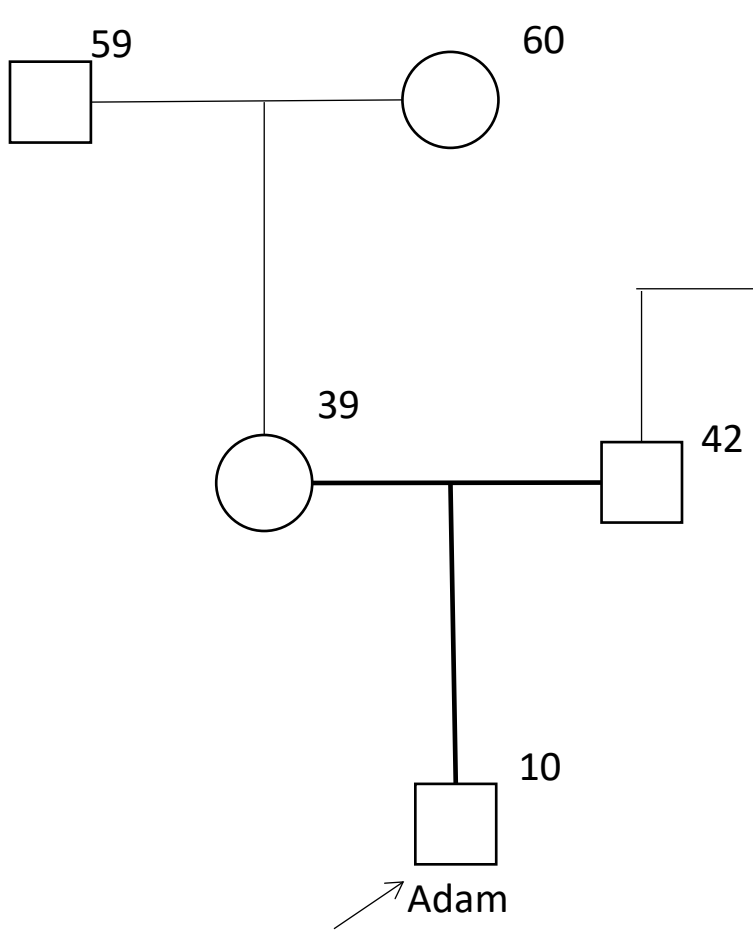
- Clarification of HCM diagnosis for individuals with borderline clinical investigations.
- Initiation of cascade testing of at-risk relatives. This can identify those at risk for HCM and those who are not at risk and would not need ongoing surveillance.
- Prognosis: positive results can indicate earlier onset of disease and worse outcomes.
- Assistance with life planning (e.g., decisions about careers, participation in competitive sports).
- Provision of relief: a negative result means one is not at higher risk to develop HCM, neither are their children

Considerations of genetic testing for HCM

Genetic testing can :

- Result in adverse psychological reaction, particularly due to potential for risk of sudden cardiac death.
- Cause uncertainty due to a genetic variant of unknown clinical significance.


Individual with HCM
Baseline clinical screening for **all first-degree** relatives by echocardiogram and ECG
Genetic testing of index case
Negative or inconclusive genetic test results
Positive results
Genetic testing for all first-degree relatives
All first-degree relatives periodic clinical screening*
Positive results
(True) Negative results
Discharge from screening (if normal baseline)
Individualized management and risk stratification
Re-visit genetic testing, q3-5y
*Periodic clinical screening with echocardiography:
A frequency of every 1-2 years to age 20, q3-5y thereafter



For Adam to be at risk, his father would have to be affected.*

The grandfather should be offered genetic testing and his first-degree relatives offered baseline screening.

All HCM surveillance recommendations are for first-degree relatives. There are none for second-degree relatives.

Hypertrophic cardiomyopathy

Exercise:

- Traditionally there have been restrictions
- Current evidence shows potential benefit of moderate exercise for stable HCM patients
- Competitive sports and vigorous exercise should be discussed with an expert in HCM

How do I care for my patients with HCM?

1. Identify red flags for a personal and family history of HCM
2. Refer affected individuals with HCM to cardiology and genetics for assessment and testing
3. Refer first-degree relatives of those affected by HCM to cardiology and genetics if a causative genetic variant is known

    Second-, third- and more distant relatives can be referred for genetic testing if a causative genetic variant is known and the intervening relative(s) is unable/unwilling to have testing

Case 2

Beth, 35yo female, BMI >30

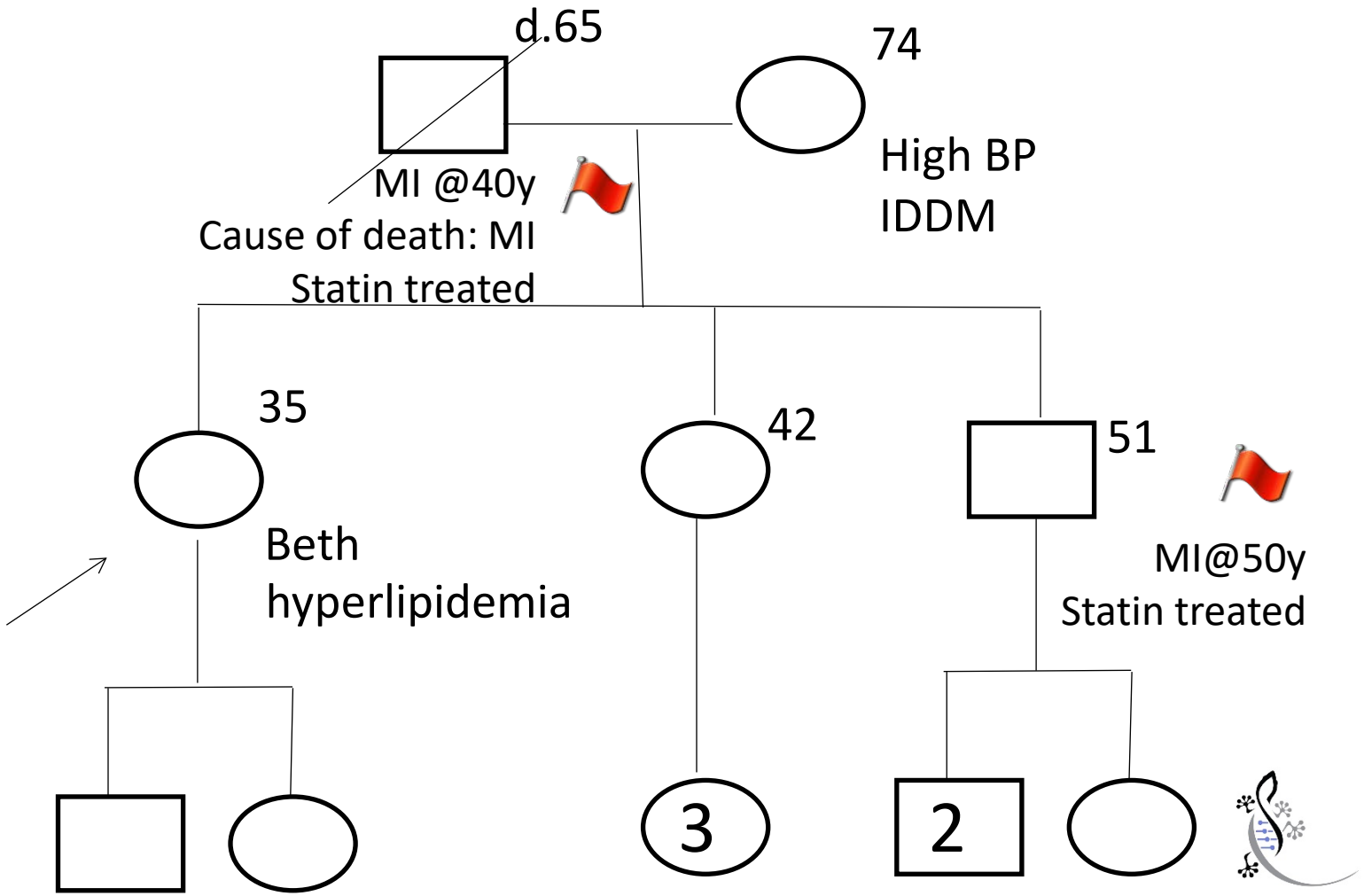
As part of a weight-loss clinic work up LDL-C was found to be 6.9mmol/L

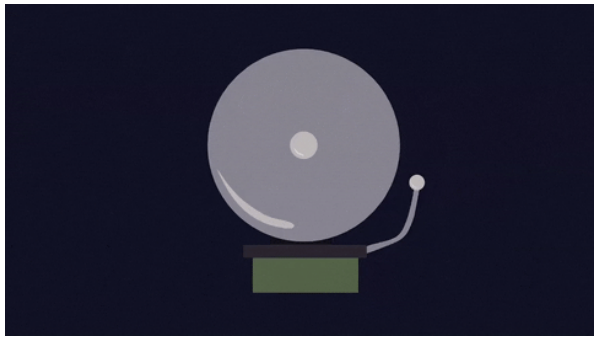
You prescribed a statin

On follow up LDL-C was 7.2mmol/L

Patient admits being non-compliant, wishes to treat hyperlipidemia with natural, lifestyle changes e.g. weight-loss, plant-based diet, exercise

You explore the family history


d.65
74
MI @40y
High BP
IDDM
Cause of death: MI
Statin treated
35
42
51
Beth
hyperlipidemia
MI@50y
Statin treated
3
2



92

dementia

stroke @50s

d.65

74

75

MI @40s

MI @40y

Cause of death: MI

Statin treated

High BP

IDDM

45

2

Statin treated

35

42

51

MI@50y

Statin treated

Beth

hyperlipidemia

3

2

Familial hypercholesterolemia (FH)

About 1 in 250 Canadians affected, but it is significantly underdiagnosed (*<10%*) and the clinical presentation is often an acute coronary syndrome

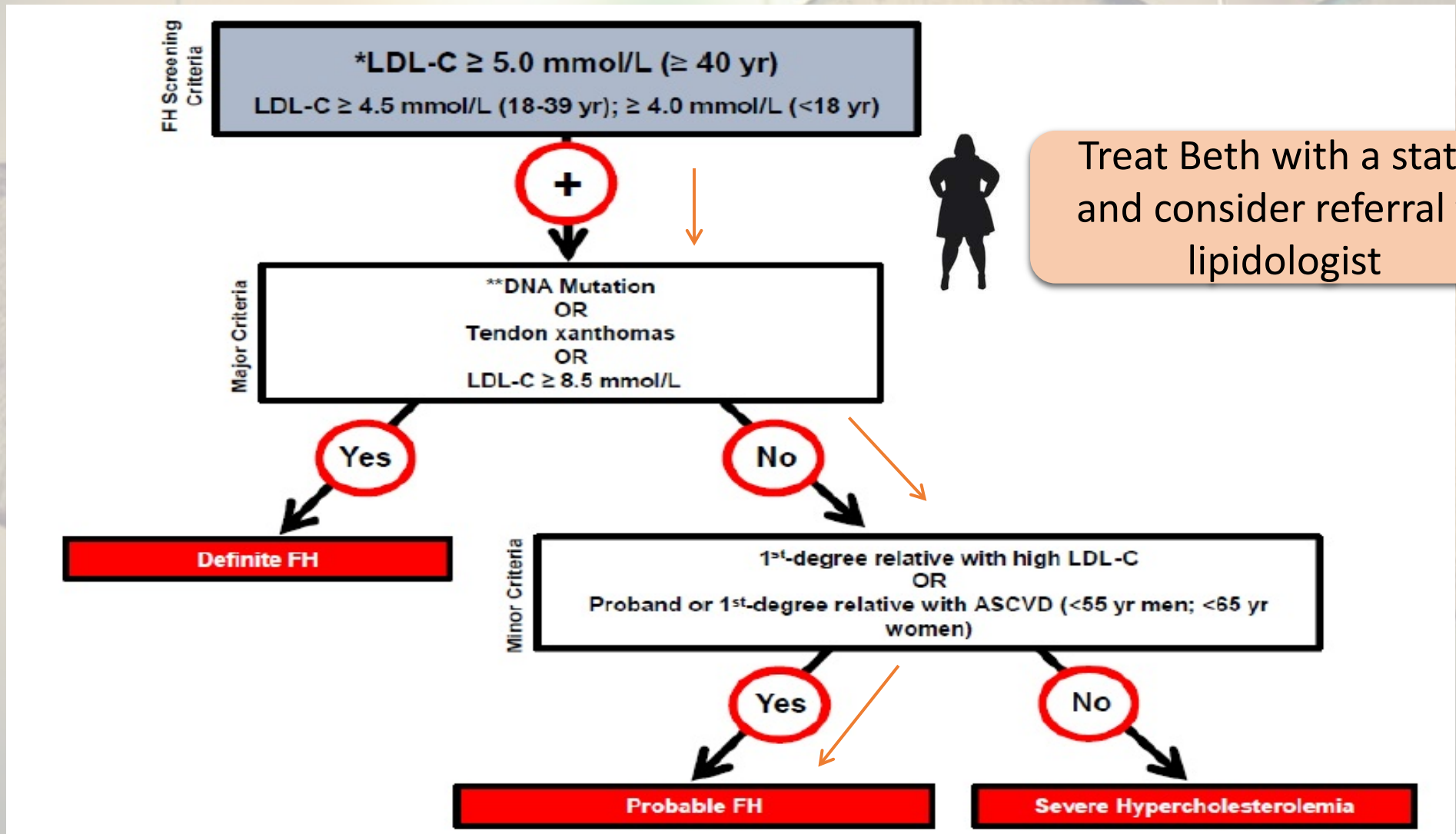
Autosomal dominant disorder resulting in 6-22x increased risk of premature cardiovascular disease (CVD) and death

Early diagnosis and treatment (statin+) can normalize life expectancy

Key features of FH are:

- elevated LDL-C ≥5 mmol/L
- early onset CVD (<55 years in men, <65 years in women)
- cholesterol deposition in the tendons (xanthomata) and/or around the eyes (xanthelasma)
- arcus cornealis with onset <45 years
- family history of early onset CVD or hyperlipidemia requiring treatment

Diagnosis | Management | Screening | Genetics

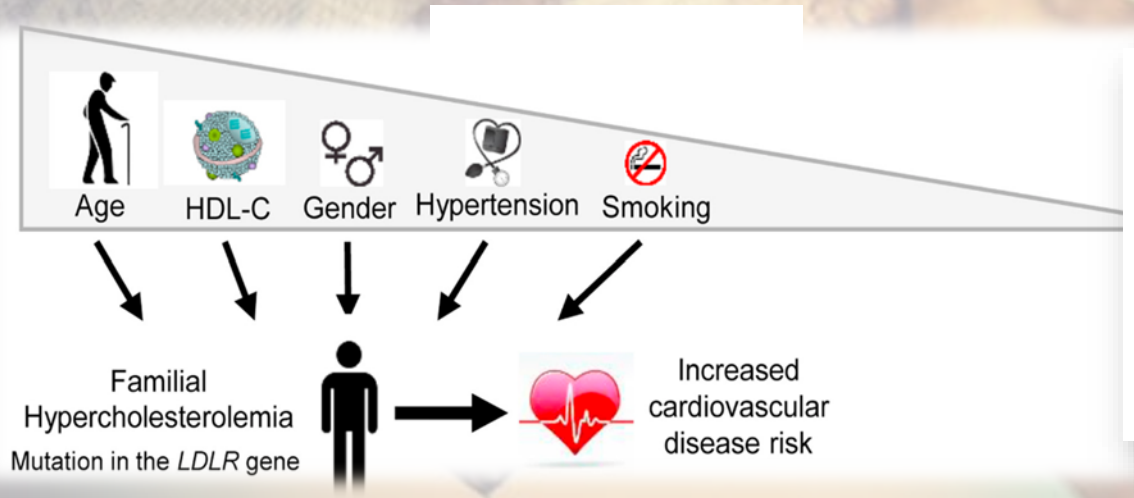


Treat Beth with a statin and consider referral to lipidologist

ASCVD: atherosclerotic cardiovascular disease

Diagnosis | **Management** | Screening | Genetics

Framingham risk score



FH-SCORE to predict CV events in FH

Pauquette 2017 J Clin Lipidol

FH Canada
Familial Hypercholesterolemia

CardioRisk Calculator ™

CardioRisk Calculator ™ simplifies cardiovascular risk stratification and is a Canadian dyslipidemia guidelines application.

www.circl.ubc.ca/cardiorisk-calculator.html

What is the most appropriate first-line pharmacologic treatment for patients with familial hypercholesterolemia?

a) Bile acid sequestrants
b) Statins
c) Fibrates
d) Niacin
e) Ezetimibe

What is the most appropriate first-line pharmacologic treatment for patients with familial hypercholesterolemia?

a) Bile acid sequestrants
b) Statins
c) Fibrates
d) Niacin
e) Ezetimibe

Diagnosis | Management | Screening | Genetics

The CCS recommends for those with FH, a >50% reduction of LDL-C (untreated) from baseline beginning at age 18 as primary prevention.

Statins are the treatment of choice for FH

Non-fasting lipid profiles should be used to monitor treatment in those whose treatment is stable

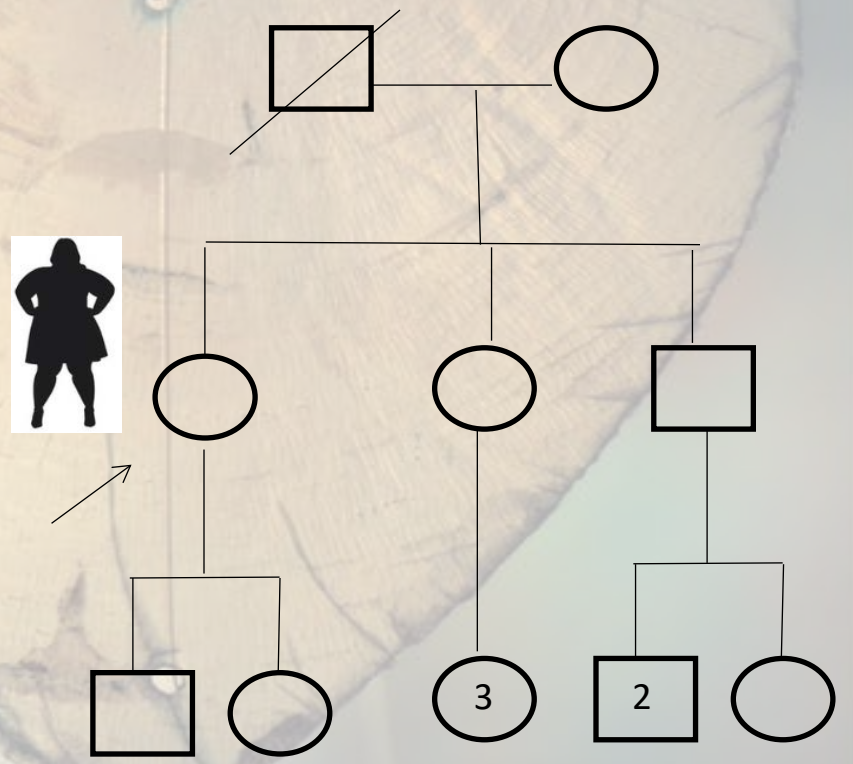
Statin-treated patients with FH have cardiovascular outcomes similar to an age- and sex-matched population without FH

Canadian Cardiovascular Society 2018

Cascade screening by lipid levels of first-degree relatives of individuals with FH

The most cost-effective approach for identification of new FH cases is cascade screening of family members.

This reduces the average age at which an individual is diagnosed and results in an increased number of individuals who are treated with statins and have subsequent lowered lipid levels.



Diagnosis | Management | Screening | Genetics

There remains controversy as to whether screening in childhood for FH should be implemented.

The CCS and Canadian Pediatric Cardiology Association recommend universal lipid screening be performed after 2 years of age within the first decade of life.

Selective screening at anytime can be considered when there is a positive family history of premature CVD or dyslipidemia, or other cardiovascular risk factors.

The ideal age to begin treatment is between ages 8-12 years based on current randomized control trials.

Pharmacological treatment can be considered, incorporating clinical judgement, family and patient preferences.

Why consider genetic testing?

If a causative genetic variant is identified, results are useful to:

- complement the clinical diagnosis and increase compliance with treatment
- enable cascade testing of family members
  - Those who are negative for the familial genetic variant are not at increased risk for hypercholesterolemia
- provide risk stratification
  - Individuals with FH-causing variants in the *LDLR, APOB,* or *PCSK9* genes are at a 5- to 22-fold increased risk of atherosclerotic cardiovascular disease

Genetic testing is publicly funded in all provinces except BC

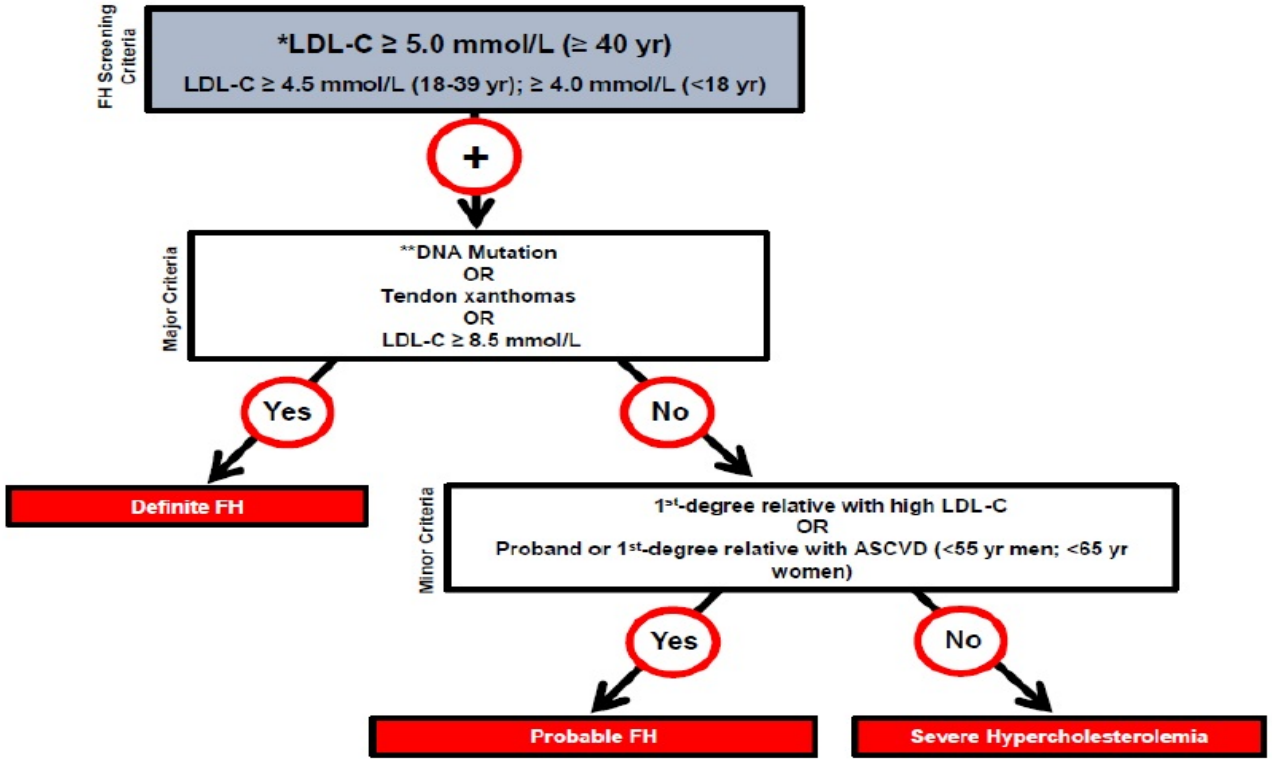
Can be ordered by family physicians in ON and QC.

Case 2

Treat Beth with a statin and consider referral to a lipidologist

Initiate cascade lipid screening of first-degree relatives

Offer and order genetic testing where available. Private pay testing may be an option.



How do I care for my patients with FH?

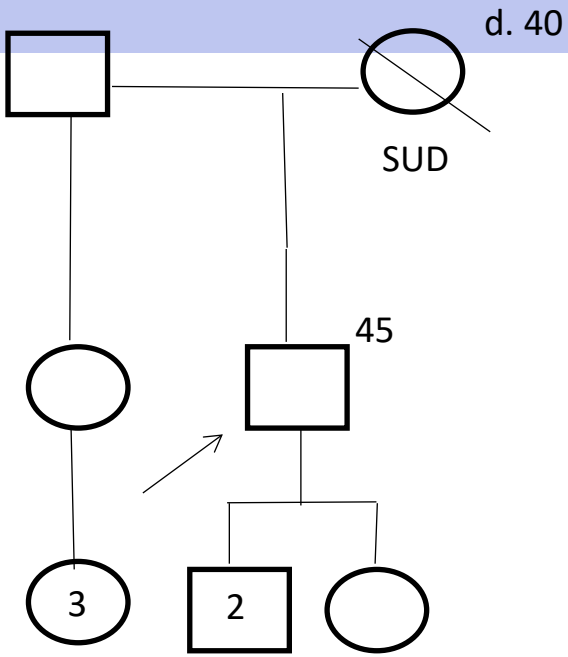
1. Identify those with probable/definite FH and consider referral to lipidologist
2. Manage LDL-C levels with statins
3. Offer and order genetic testing where available
4. Coordinate cascade screening by lipids or genetic testing

Case 3

You are seeing 45yo, Carl, to review results of an echocardiogram ordered following a visit to the ER with reported chest pain.

The report indicated mild thoracic aortic dilation

You are already aware Carl reported his mother passed away at age 40 from sudden cardiac death



Heritable Thoracic Aortic Disease (HTAD)

Key definitions:

Dilation: when the diameter of the aorta exceeds the norms for a given age and body size. Reported as borderline, mild, moderate or severe on imaging.

Aneurysm: a dilation >50% larger than the blood vessel should be. All aneurysms are dilations, however not every dilation will reach the size of an aneurysm.

Dissection: a rip or tear in the inner lining of a blood vessel.

Degenerative: an aneurysm or dilation caused by the deterioration of a blood vessel over time, associated with risk factors such as high blood pressure and age.

What about other aneurysms?

- Abdominal aortic aneurysm (AAA) is more common in the general population and is much more strongly associated with traditional risk factors for coronary artery disease.
- Intracranial aneurysm (IA) occur in approximately 2-5% of adults. The most common genetic association is with Autosomal Dominant Polycystic Kidney Disease
- AAA and IA are less likely to be associated with a strong genetic predisposition than TAA, however, some conditions predisposing to HTAD can also predispose to AAA or IA.
- In this context, aneurysms outside of the thoracic aorta are still considered pertinent *if there is a family or personal history of TAA*.

Recommendations on screening for abdominal aortic aneurysm in primary care. CMAJ 2017.

Thoracic Aortic Disease

Most thoracic aortic aneurysms (TAA) are degenerative and are primarily caused by age and hypertension.

TAA may also have infectious/inflammatory and traumatic etiologies.

Heritable Thoracic Aortic Disease (HTAD)

HTAD accounts for **~20-25%** of all thoracic aortic aneurysms and dissections

Most individuals with HTAD do not have additional associated features **(non-syndromic**), although some will e.g. Marfan syndrome, vascular Ehlers-Danlos Syndrome

HTAD presents at a **younger** age and is **more aggressive** than other thoracic aortic aneurysms

Appropriate recognition of HTAD allows initiation of imaging surveillance in at-risk relatives.

Heritable Thoracic Aortic Disease (HTAD)

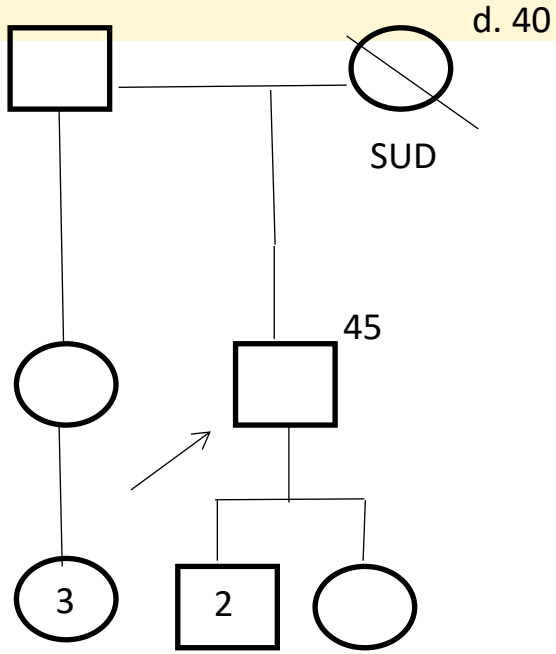
CONSIDER REFERRAL FOR GENETIC ASSESSMENT FOR THOSE WITH:

- ☐ Thoracic aortic dilation reported on imaging as mild or greater, at age <50y or <60y in the absence of hypertension
- ☐ Thoracic aortic dissection at age <60y or <70y in the absence of hypertension
- ☐ Thoracic aortic dilation at any age in the presence of any of the following family histories in a 1st or 2nd degree relative:
  - ☐ Thoracic aortic aneurysm (TAA) or thoracic aortic dissection
  - ☐ Sudden cardiac death age at <50y without a confirmed alternate etiology
- ☐ Personal or family history of thoracic aortic dilation or TAA and/or features that suggest an underlying syndromic condition, such as:
  - ☐ Tall for family
  - ☐ Ectopia lentis (lens dislocation
  - ☐ Spontaneous pneumothorax (particularly if recurrent)
  - ☐ Hypertelorism (wide-spaced eyes)
  - ☐ Bifid uvula
  - ☐ Hollow organ rupture
  - ☐ Spontaneous tendon rupture
  - ☐ Large and unprovoked bruising (prior to anti-coagulation)
  - ☐ Very translucent skin
  - ☐ Pectus carinatum or significant pectus excavatum
  - ☐ Scoliosis requiring bracing or surgery
  - ☐ Significant varicose veins at a young age
- ☐ 1st or 2nd degree relative in whom a pathogenic or likely pathogenic variant in one of the HTAD genes has been identified
  - referral of 3rd degree relatives can be considered when intervening relatives are not available or decline testing

Case 3

Refer Carl to cardiology and/genetics for an assessment of Heritable Thoracic Aortic Disease

Include echocardiogram report, medical history family history (try to take more of the maternal history), patient could obtain coroner's report if mother had autopsy



In heritable thoracic aortic disease (HTAD), which of the following is the most important reason for genetic testing of family members?

a) To predict the exact age of aneurysm formation
b) To identify individuals at risk before symptoms develop
c) To determine if surgery is immediately necessary
d) To rule out other cardiovascular diseases
e) To assess risk of aneurysm rupture

In heritable thoracic aortic disease, which of the following is the most important reason for genetic testing of family members?

a) To predict the exact age of aneurysm formation
b) To identify individuals at risk before symptoms develop
c) To determine if surgery is immediately necessary
d) To rule out other cardiovascular diseases
e) To assess risk of aneurysm rupture

Why offer genetic testing for HTAD?

Genetic testing is ordered by a specialist and is panel-based (~19 genes)

A positive genetic test result can help:

- guide pharmacotherapy
- determine vascular regions which require ongoing imaging surveillance
- influence surgical threshold
- allow for cascade testing of at-risk relatives

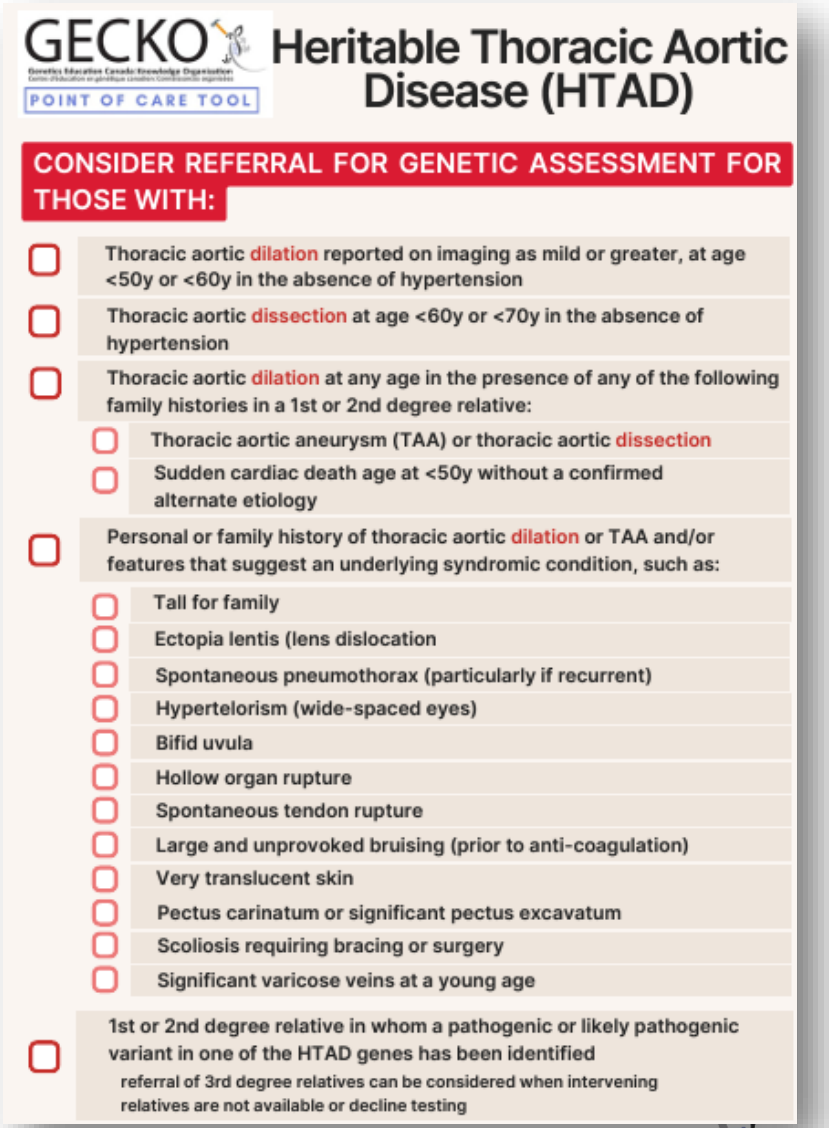
In most families with HTAD (80%), genetic testing does NOT identify the responsible genetic variant

→ Negative testing does not exclude HTAD and at-risk relatives would still need ongoing imaging surveillance

How do I care for my patients with HTAD?

1. Identify patients who would benefit from referral to an aortopathy clinic/cardiologist and genetics expert for assessment and testing

2. Initiate referrals to facilitate cascade screening of relatives through an aortopathy and/or a genetics clinic



GECKO
POINT OF CARE TOOL

Heritable Thoracic Aortic Disease (HTAD)

CONSIDER REFERRAL FOR GENETIC ASSESSMENT FOR THOSE WITH:

- ☐ Thoracic aortic dilation reported on imaging as mild or greater, at age <50y or <60y in the absence of hypertension
- ☐ Thoracic aortic dissection at age <60y or <70y in the absence of hypertension
- ☐ Thoracic aortic dilation at any age in the presence of any of the following family histories in a 1st or 2nd degree relative:
  - ☐ Thoracic aortic aneurysm (TAA) or thoracic aortic dissection
  - ☐ Sudden cardiac death age at <50y without a confirmed alternate etiology
- ☐ Personal or family history of thoracic aortic dilation or TAA and/or features that suggest an underlying syndromic condition, such as:
  - ☐ Tall for family
  - ☐ Ectopia lentis (lens dislocation
  - ☐ Spontaneous pneumothorax (particularly if recurrent)
  - ☐ Hypertelorism (wide-spaced eyes)
  - ☐ Bifid uvula
  - ☐ Hollow organ rupture
  - ☐ Spontaneous tendon rupture
  - ☐ Large and unprovoked bruising (prior to anti-coagulation)
  - ☐ Very translucent skin
  - ☐ Pectus carinatum or significant pectus excavatum
  - ☐ Scoliosis requiring bracing or surgery
  - ☐ Significant varicose veins at a young age
- ☐ 1st or 2nd degree relative in whom a pathogenic or likely pathogenic variant in one of the HTAD genes has been identified
  referral of 3rd degree relatives can be considered when intervening relatives are not available or decline testing

Genetic test results

☆The best person to begin testing is the youngest, affected relative.

Testing is done with a gene panel where several genes are analyzed at once. Genes are curated based on the indication for testing.

Proportion of patients that will have a positive result

Hypertrophic cardiomyopathy	35-60%
Familial hypercholesterolemia	30-80% *(LDLR, APOB,* or *PCSK9)*
Heritable Thoracic Aortic Disease	20%

Genetic test results: Positive

Positive: causative pathogenic or likely pathogenic variant detected in a condition related gene

This confirms a genetic etiology and a diagnosis of the suspected condition.

Result may be used to guide management, screening, risk stratification, etc.

Genetic testing can be offered to relatives for the familial gene variant. Each first-degree relative has a 50% change to inherit the familial variant and should be offered testing.

- Those identified to be at-risk can be appropriately managed, screened, referred.
- Those identified to not be at-risk can be reassured they are at population risk.

Genetic test results: Negative

Uninformative: no clinically significant genetic variants detected

- A genetic etiology is neither confirmed nor ruled out.
- Genetic testing cannot be offered to unaffected relatives. First-degree relatives may need increased screening.
- Over time, a better test may become available and re-referring to genetics can be considered in approximately 3-5 years.

True negative: the familial pathogenic/ likely pathogenic variant is not detected

- This is where genetic testing is offered to an unaffected relative after the causative pathogenic gene variant has been identified.
- The individual and their children do not have the familial condition and is not at increased risk for the condition.

Genetic test results: Variant of uncertain significance

VUS: variant in condition-related gene is detected, but there is insufficient evidence to determine if it is truly associated with disease. (It is possible for more than one VUS to be detected.)

- This is not to be treated as a positive result.
- The genetics team may offer testing to other relatives to see if the variant is present in those with disease and absent in unaffected relatives (segregation study).
- Over time, additional information about the variant may become available. 10-15% of variants are reclassified as benign or pathogenic. Consider re-referral to genetics in approximately 3-5 years.

Many cardiac conditions have a hereditary component

Presentations that point to possible hereditary cardiac conditions and prompt further investigation include:

- Sudden death < 40 years, including SIDS
- Syncope or pre-syncope with exercise, intense emotional, stressful, or startling events
- Known family history
- Seizures/Drowning/Unexplained single MVA
- Cardiac device at a young age (ICD, pacemaker)
- Cardiac disease *without* usual risk factors

Identification of at-risk family members allows for early intervention and lifesaving surveillance and management

resources www.geneticseducation.ca

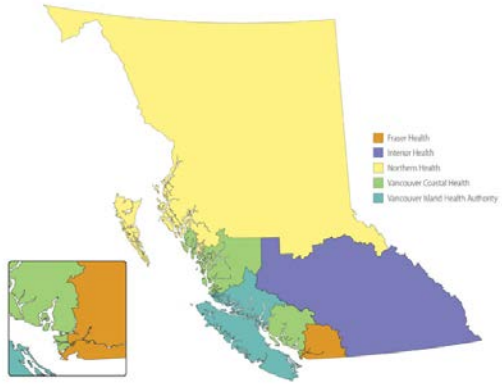
Resources for clinicians > Cardiogenomics

GECKO
Genetics Education Canada: Knowledge Organization
Centre d'éducation en génétique canadien: Connaissances organisées
POINT OF CARE TOOL

GECKO
Genetics Education Canada: Knowledge Organizat
Centre d'éducation en génétique canadien: Connaissances organise
on the run

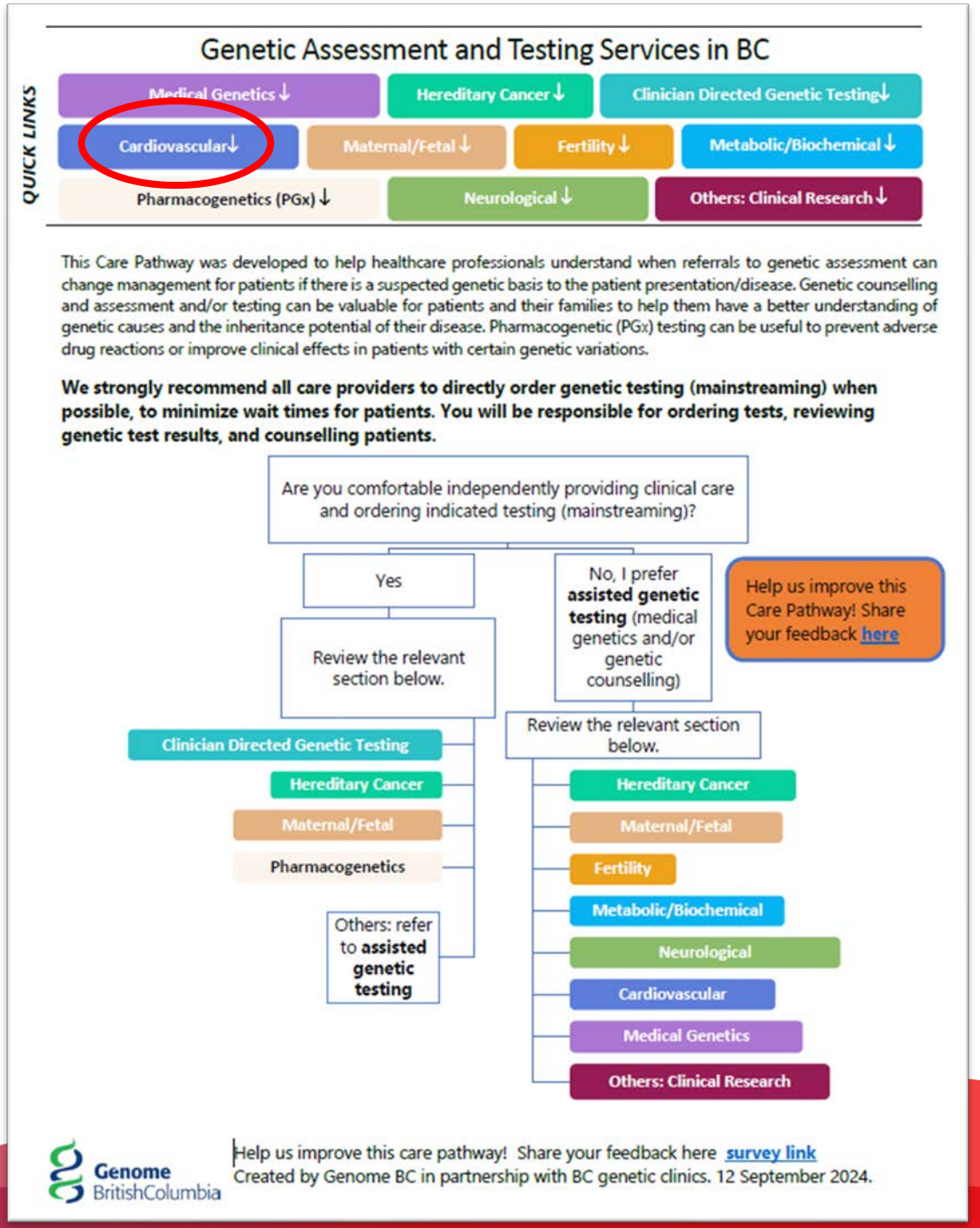
GECKO
Genetics Education Canada: Knowledge Organization
Centre d'éducation en génétique canadien: Connaissances organisées
DEEP DIVE

Pathways™



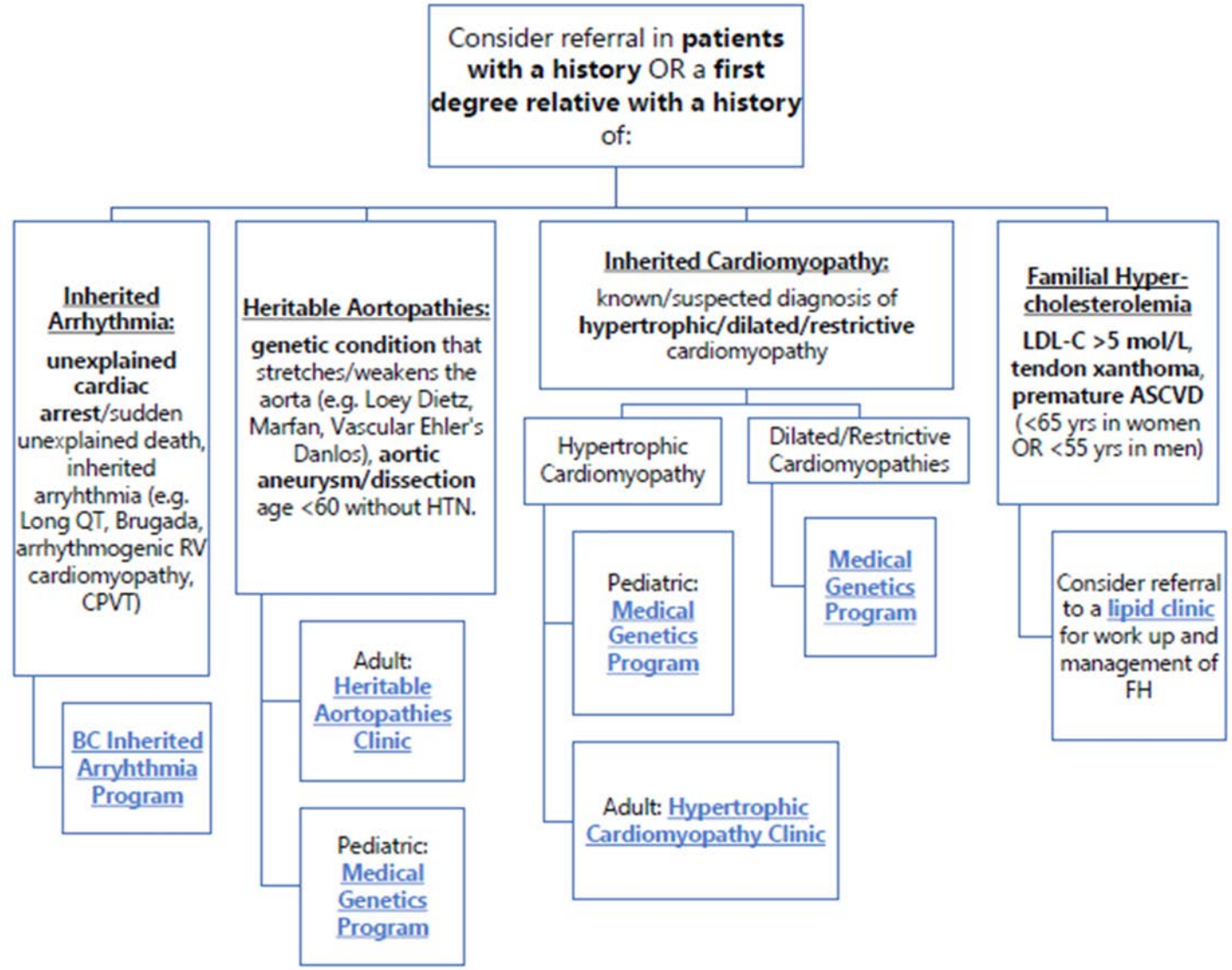
Genetic Assessment and Testing Services - Enhanced Care Pathway

Genetic clinical services available for BC residents in one central location.



Cardiac Services BC
Provincial Health Services Authority

Cardiovascular



Questions

Morrison@cheo.on.ca

Kirsten.Bartels@phsa.ca

June.Carroll@sinaihealth.ca

Thank you!

Please fill out your session evaluation now!

#myfmf

 FamilyMedicineForum

 FamilyMedForum

 FamilyMedForum

HCM

Canadian Cardiovascular Society (2024)
https://onlinecjc.ca/article/S0828-282X(24)00438-0/fulltext

https://geneticseducation.ca/resources-for-clinicians/cardiogenomics/hypertrophic-cardiomyopathy

FH

Canadian Cardiovascular Society (2018)
https://onlinecjc.ca/article/S0828-282X(18)31171-1/fulltext

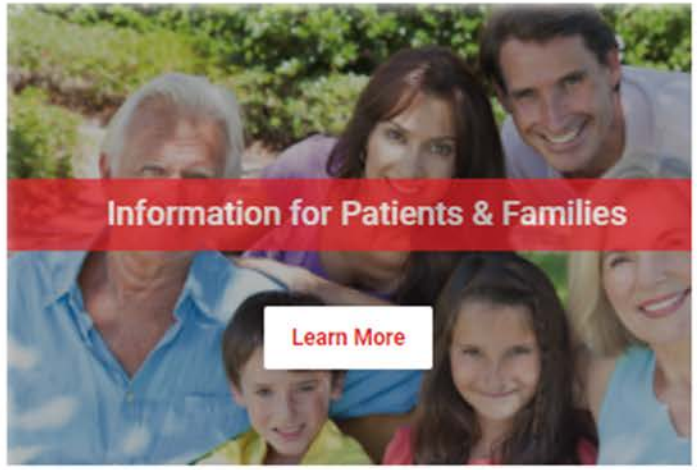
https://geneticseducation.ca/resources-for-clinicians/cardiogenomics/familial-hypercholesterolemia

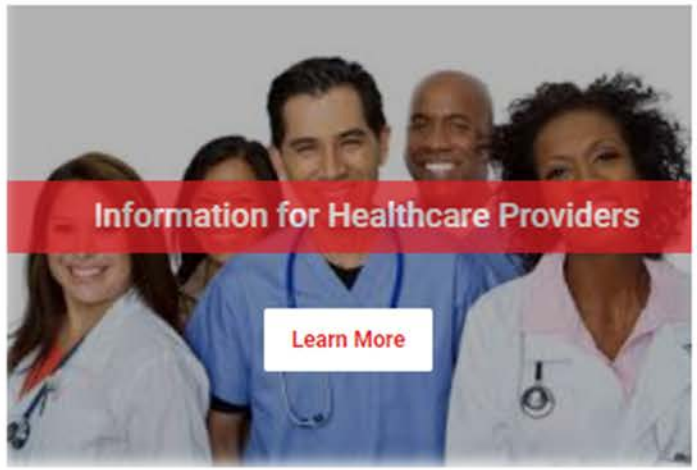
HTAD

https://geneticseducation.ca/resources-for-clinicians/cardiogenomics/htad-landing-page

resources www.sads.ca

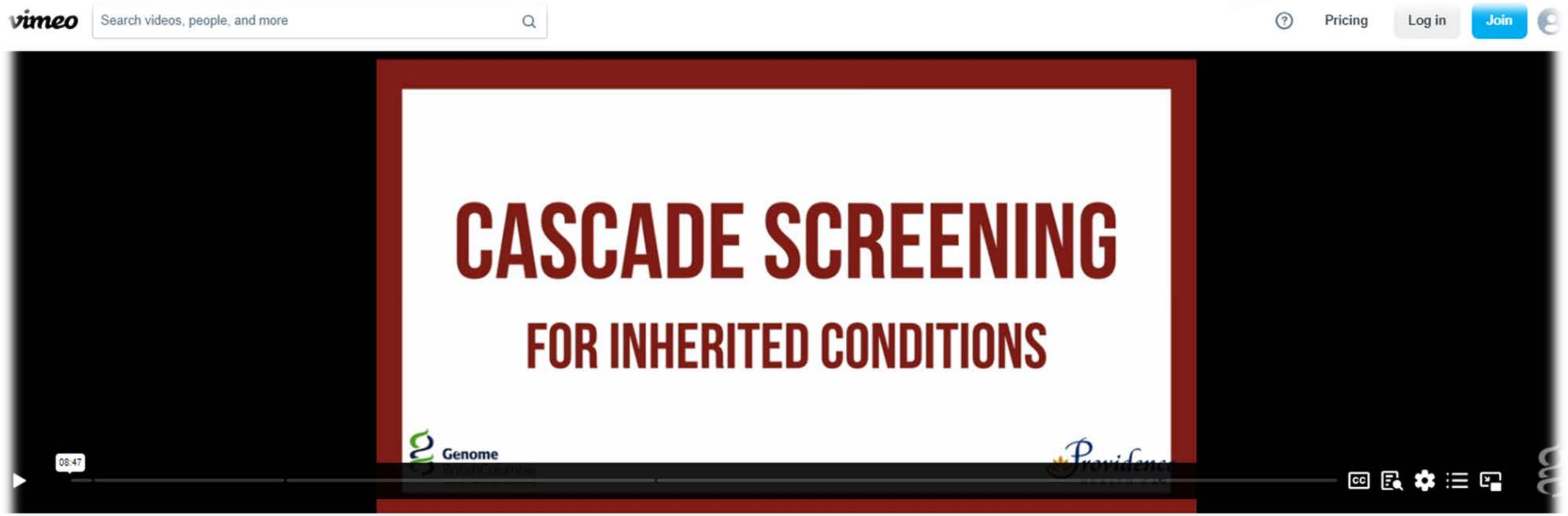
The Canadian Sudden Arrhythmia Death Syndromes Foundation





resources https://vimeo.com/826986524/7969433f18

Genome BC



resources www.hiro.heartsinrhythm.ca

Heart in Rhythm Organization

resources https://www.genomebc.ca/fh-resources

Genome BC

Familial Hypercholesterolemia Resources

To support physicians with integrating genetic testing at the point of care, Genome BC, as part of their Genomics Education for Health

resources https://www.fhcanada.net/

FH Canada

HF Canada
Hypercholestérolémie Familiale

FH Canada
Familial Hypercholesterolemia

Patients

Français English

Mission, Vision, Goal

The MISSION of the Canadian FH Registry is to bring together a multi-disciplinary group of physicians, basic and clinical researchers to improve the delivery of care to patients with severe lipoprotein disorders, especially FH, and to foster collaborative research.

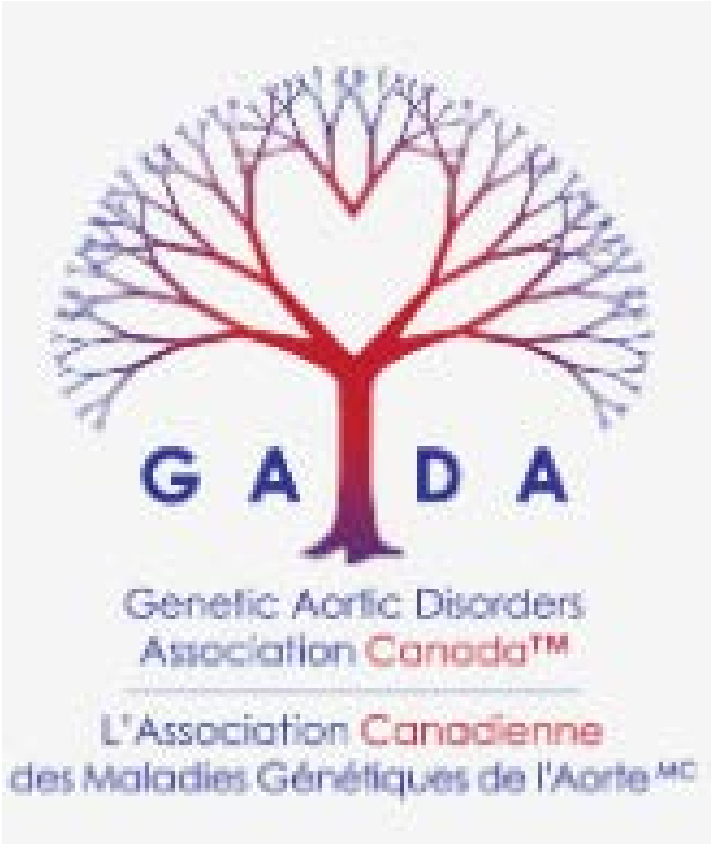
Our VISION is to create a Canada-wide network of academic clinics, integrating lipid specialists, endocrinologists and cardiologists to treat patients with the highest standard of care and to create a collaborative research environment. Using a "hub and spoke" model, the registry will be extended in various communities to link primary care physicians with provincial academic centers.

The GOALS are to improve care to patients with FH and to reduce cardiovascular disease in this population at high risk.

Physicians

resources https://www.gadacanada.ca/clinics-across-canada-1

Canadian Aortopathy Clinics via Genetic Aortic Disorders Association Canada



resources https://www.ottawacvgenetics.ca/

Ottawa CardioVascular Genetics