HF Canada
Hypercholestérolémie Familiale

FH Canada
Familial Hypercholesterolemia

Canadian Familial Hypercholesterolemia Registry
Régistre Canadien d'hypercholestérolémie familiale

Aim of FH Canada registry

- **The aim of the FH registry is to improve the detection and management of individuals and families with FH in Canada. Rare diseases of lipoprotein metabolism are also included (SMASH initiative)**

- **Initiated at University of British Columbia and became national in 2014.**

- **Over 200 clinicians and scientists in 19 academic centers across Canada form the "hubs" of FH Canada.**

Clinicaltrials.gov: NCT02009345

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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.

FH Canada Registry “hub and spoke” model

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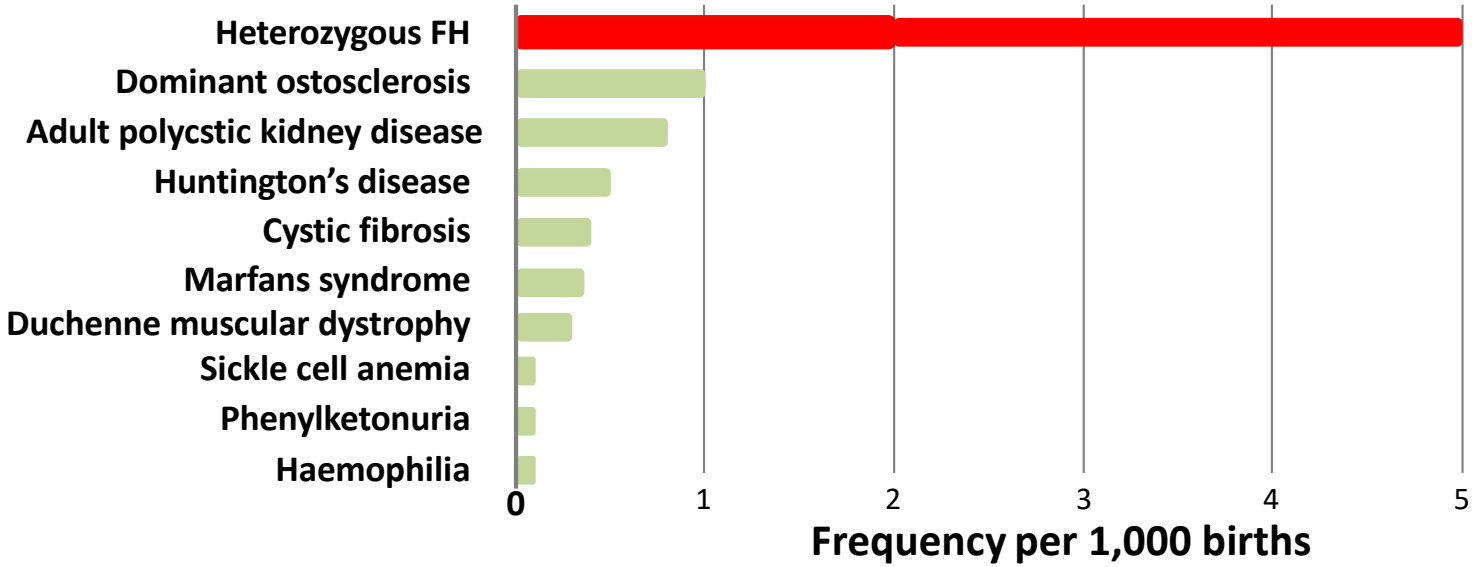
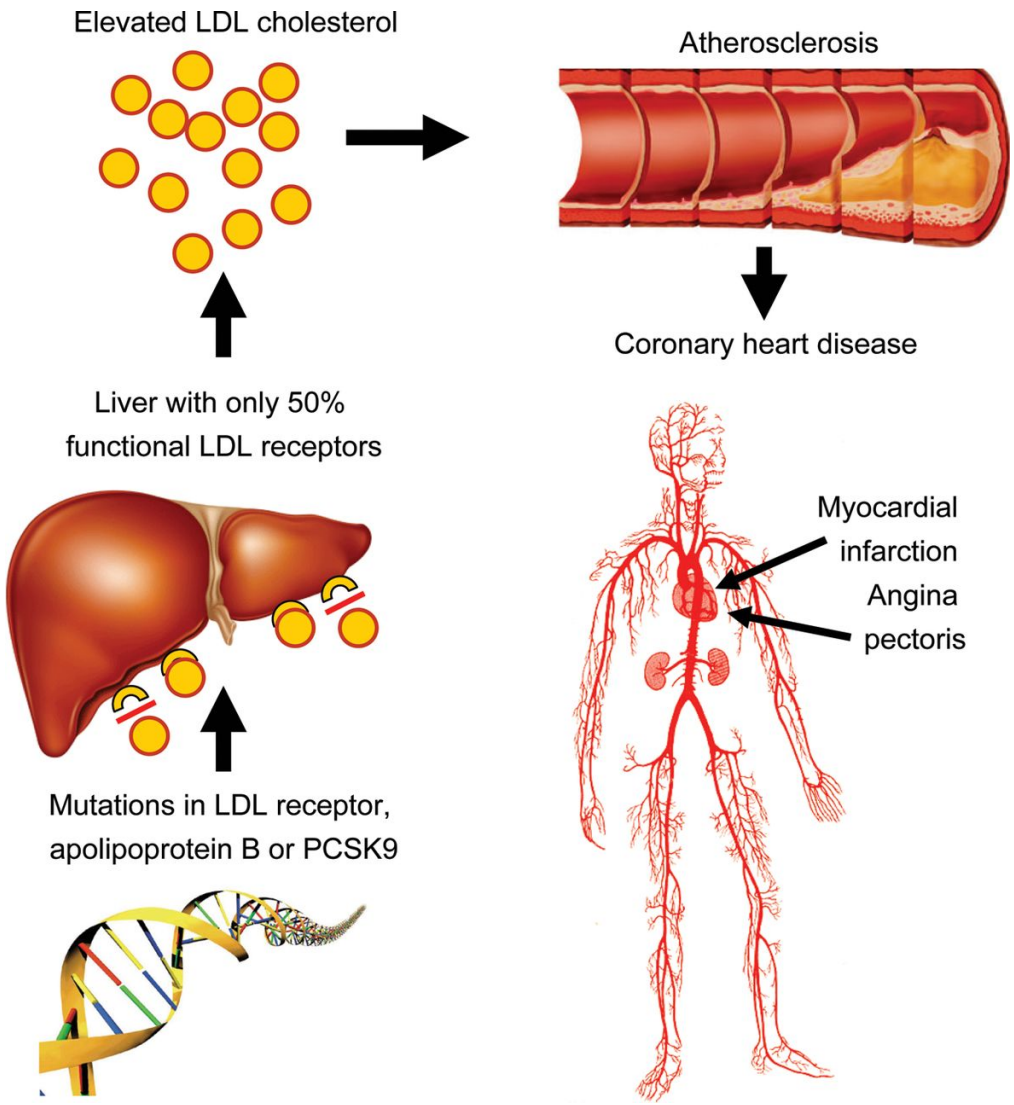
FH is One of the Most Common of Inherited Diseases



- Heritable, autosomal co-dominant disorder[1]
- Usually due to mutations in LDL receptor gene[2,3]
  - > 1800 mutations
  - LDLR mutation 1 in 250
  - ~ 143,000 patients in Canada, with less than 10% of patients diagnosed

1.Marais AD. Clin Biochem Rev. 2004;25:49-68.
2.Mahley RW, et al. In: Kronenberg: Williams Textbook of Endocrinology. 2008.
3.Rader DJ, et al. J Clin Invest. 2003;111:1795-1803.

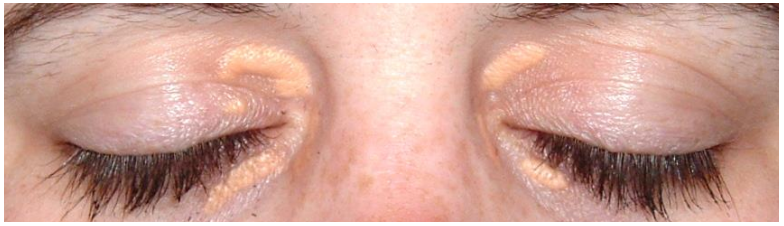
Pathophysiology of HeFH



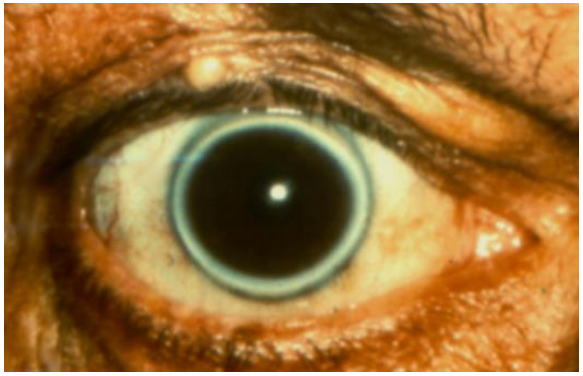
Nordestgaard B G et al. Eur Heart J 2013;34:3478-3490

European Heart Journal

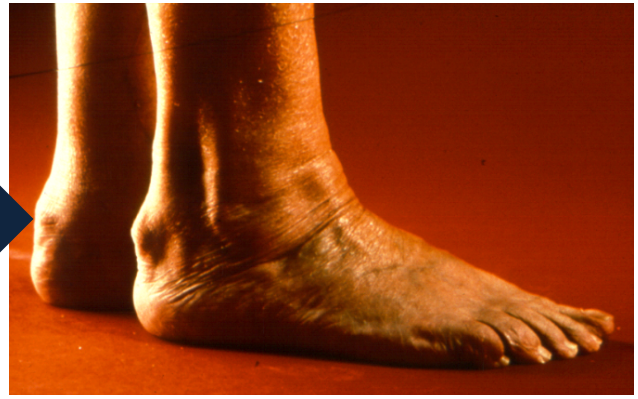
Clinical manifestations



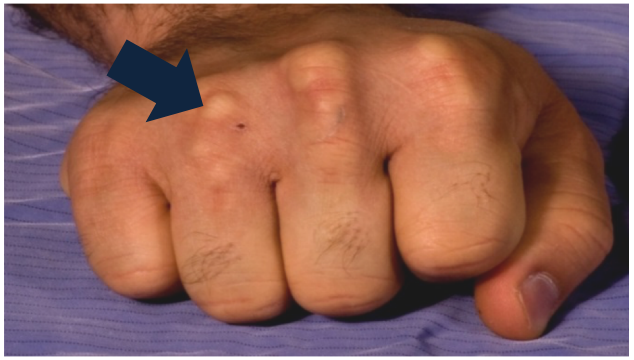
Bilateral xanthelasma



Arcus Cornae



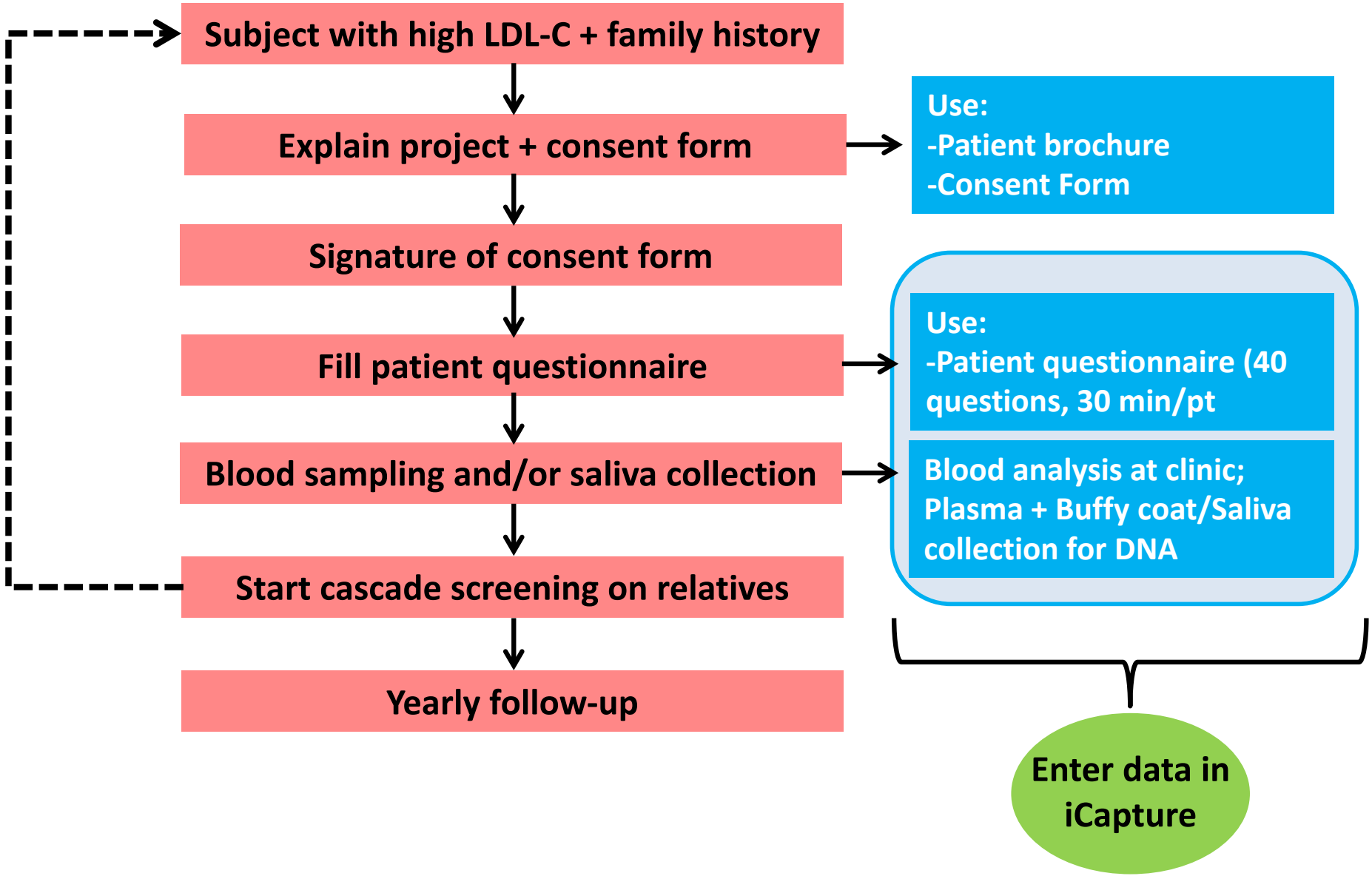
Xanthomas within the Achilles tendons



Xanthoma within extensor tendon of the hand

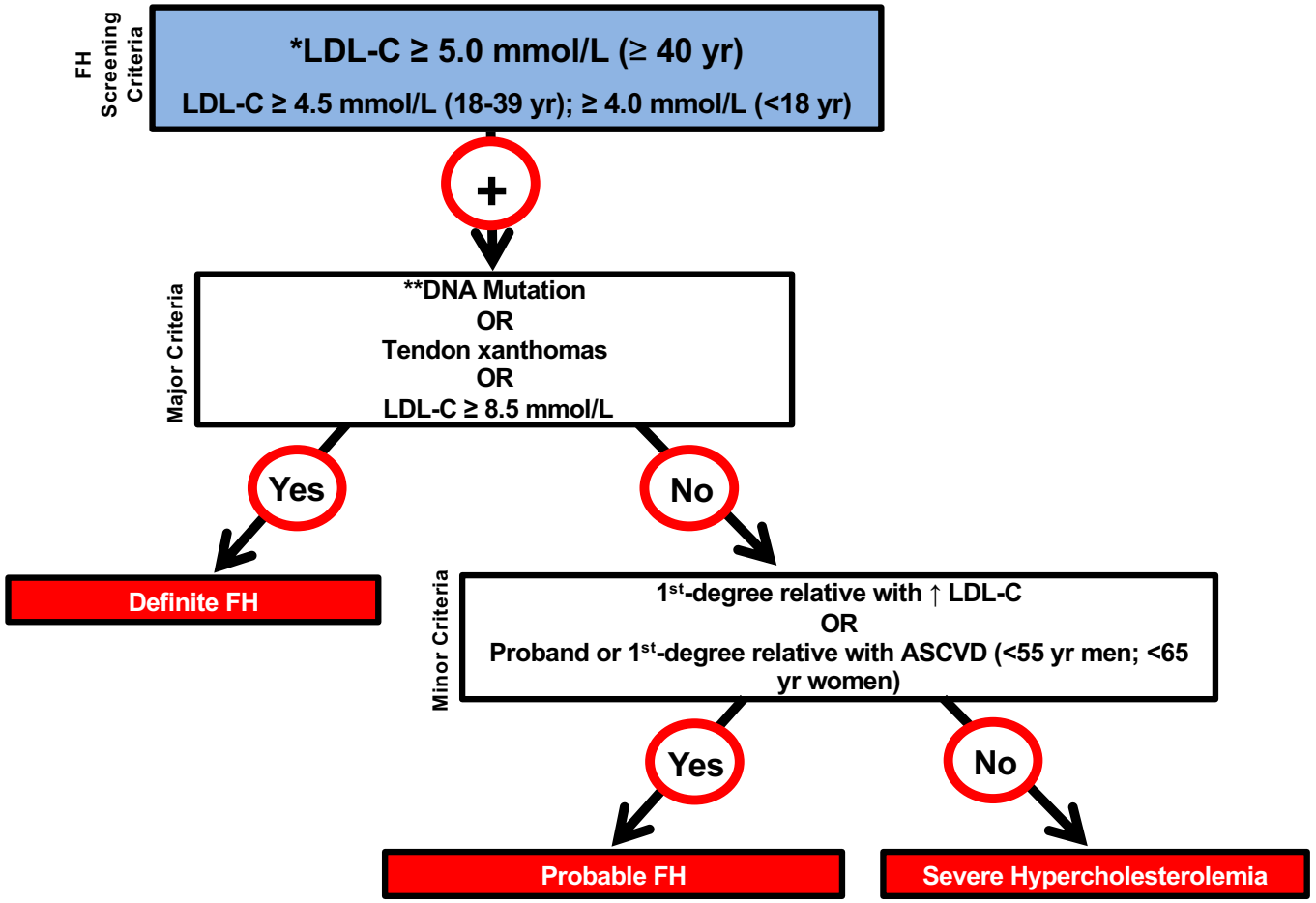
Genest J, Hegele RA, Bergeron J, Brophy J, Carpentier A, Couture P, et al. Canadian Cardiovascular Society position statement on familial hypercholesterolemia. The Canadian journal of cardiology. 2014;30(12):1471-81.

Overview: What does the study involve?



Inclusion criteria: Canadian definition

Any patient with a clinical diagnostic criteria for FH:



*** Secondary causes ruled out (nephrotic syndrome, obstructive jaundice, hypothyroidism, drugs, other).**

**Mutation in LDL-r, ApoB or PCSK9; Presence of a DNA causing mutation in a proband is sufficient for a diagnosis of FH.

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Data collected from patient questionnaire

***Mainly "Yes/No" answers, max 30 min/patient**

Section 1: Patient Data

- Demographics (name, address, birth date, gender, self-reported ethnicity, consent form signed + date, preferred method of contact)
- Family doctor contact info

Section 2: Past medical Exam

- Family history (familial history of CVD and high LDL-C in 1st-degree relatives)
- Smoking status (non-, ex or smoker, nb years, nb cigarettes/day)
- Medical history (hypertension, DM, CAD as in MI, Angina, etc)
- Surgical history (PCI, CABG, arterial revascularisation)

Section 3: Physical exam

- Standard measurements (weight, height, blood pressure)
- Physical signs of FH (corneal arcus <45 years old, xanthelasma, xanthomas)

Section 4: Medication data

- Lipid-lowering medication (statin, ezetimibe, etc with dose and frequency, statin intolerance)
- Non lipid-lowering medication (prescribed or over-the-counter with dose and frequency)

Section 5: Lab data entry

- Date of analysis
- Fasting as Yes/No
- Known or self-reported baseline LDL-C (untreated)
- Blood glucose, Hemoglobin A1C, Total cholesterol, LDL-C, HDL-C, TG, CK, Creatinine, AST, ALT, Lipoprotein (a), ApoB

Section 6: Genetic

- DNA gene mutation(s) if known (DNA isolation Yes/No, gene + mutation name)
- SNP score if known

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Entering data in iCapture

- The James Hogg Research Centre at St-Paul's Hospital, UBC, Vancouver is providing the **iCAPTURE platform** used to capture the data from the FH Canada Registry.

- The database utilizes an Oracle backend and is firewalled and maintained in a separate non public network, and it is FDA, Health Canada, PHIA and PIPEDA compliant. All user access is logged.

- A unique identifier will be randomly assigned to each patient (0 to 999999) and only this number will be used in the de-identified national registry.

- All de-identified data from the registry will be made available for statistics on FH in Canada, for health outcomes and health economic studies which will help allow resource allocation and quality control.

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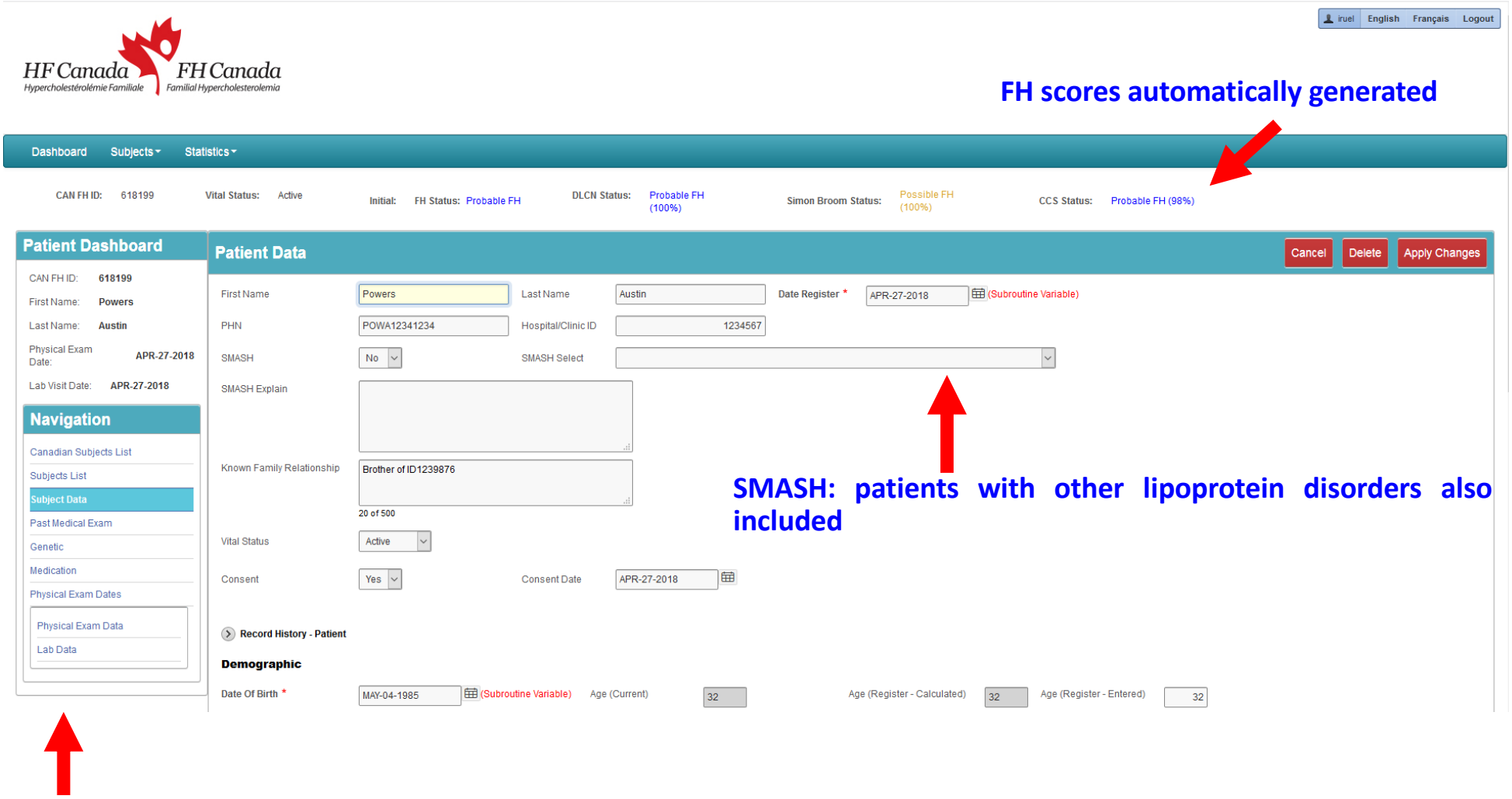
iCapture database



Data collected include demographics, family history of high LDL-C or CVD, patient's medical history, physical signs of FH, meds and lab data (blood glucose, HbA1C, total cholesterol, LDL-C, HDL-C, TG, CK, creatinine, AST, ALT, Lp(a), ApoB.

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The database has built-in algorithms to generate a score for the most common FH criteria (Simon-Broome, Dutch Lipid Clinic Network (DLCN), Canadian definition).

Simon Broome Register criteria for the clinical diagnosis of familial hypercholesterolemia (FH)

Description			Criteria
Presence of DNA mutation known to cause FH (*LDLR*, *APOB*, *PCSK9* genes)			**Definite**
LDL-C > 4.9 mmol/L in adults (> 4.0 mmol/L in children under 16yr) **or** Total cholesterol > 7.5 mmol/L in adults (> 6.7 mmol/L in children under 16yr)	+	Tendon xanthomas or evidence of these signs in first- or second-degree relative	**Definite**
LDL-C > 4.9 mmol/L in adults (> 4.0 mmol/L in children under 16yr) **or** Total cholesterol > 7.5 mmol/L in adults (> 6.7 mmol/L in children under 16yr)	+	Family history of myocardial infarction before age 50 yr in a second-degree relative or before age 60 yr in a first-degree relative **or** Family history of raised total cholesterol concentration > 7.5 mmol/L in a first- or second-degree relative or > 6.7 mmol/L in children under 16 yr	**Possible**

Adapted from *Risk of fatal coronary heart disease in familial hypercholesterolaemia. Scientific Steering Committee on behalf of the Simon Broome Register Group. BMJ 1991;303:893-6.*

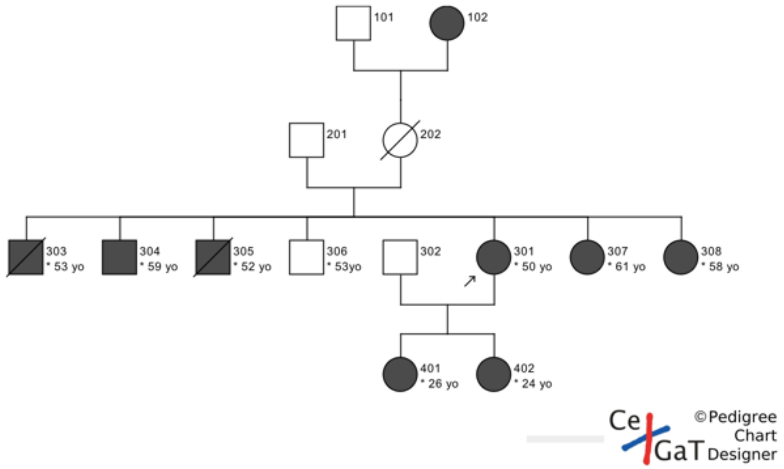
Dutch Lipid Clinic Network criteria for the clinical diagnosis of familial hypercholesterolemia (FH)	
Group 1: Family history	
• First-degree relative known with premature coronary and vascular disease (men under 55 yr, women under 60 yr) **or** • First-degree relative known with LDL-C > 95th percentile	**1 point**
• First-degree relative with tendon xanthomata and/or arcus cornealis **or** • Children under 18 yr with LDL-C > 95th percentile	**2 points**
Group 2: Clinical history	
• Patient has premature (men under 55 yr, women under 60 yr) CAD	**2 points**
• Patient has premature (men under 55 yr, women under 60 yr) cerebral or peripheral vascular disease	**1 point**
Group 3: Physical examination	
• Tendon xanthomata	**6 points**
• Corneal Arcus under 45 yr	**4 points**
Group 4: Laboratory analysis	
• LDL-C > 8.5 mmol/L	**8 points**
• LDL-C 6.5 - 8.50 mmol/L	**5 points**
• LDL-C 5.0 - 6.49 mmol/L	**3 points**
• LDL-C 4.0 - 4.99 mmol/L	**1 point**
Group 5: DNA analysis	
• Functional mutation known to cause FH	**8 points**
FH DIAGNOSIS	
• **Definite** • **Probable** • **Possible** ***The highest score per group should be applied***	**9 or more points** **6-8 points** **3-5 points**

Adapted from *World Health Organization. Familial Hypercholesterolemia - Report of a Second WHO Consultation. Geneva, Switzerland 1999.*

Increase awareness of FH: Cascade screening strategy

1- The first patients to be recruited are those with a high LDL-C already followed at the site clinic.

2- Then, family members and other undiagnosed patients (ex. siblings and cousins) are recruited from cascade screening and are referred to the nearest FH Canada participating site (www.fhcanada.net).



3- New patients are recruited with the help of the FH Canada website and the increasing awareness of FH in Canada.

What has been done so far:

1- Publication of a snapshot of the FH Canada registry - 2018

2- Characterization of the prevalence of FH (Meta-analysis)

3- Validation of a simpler definition of FH for Canadians

4- Validation of an algorithm to impute the baseline LDL-C when patient is on treatment and baseline LDL-C is unknown

5- Creation of a new FH Canada “App” - Apple and Android to ease the diagnosis of FH

6- Update of the CCS Position Statement on FH

7- Set-up the genetic testing for FH – complete DNA sequencing at MUHC.

1- Snapshot of the FH Canada registry

Atherosclerosis 277 (2018) 419–424

Contents lists available at ScienceDirect

Atherosclerosis

journal homepage: www.elsevier.com/locate/atherosclerosis

Familial hypercholesterolemia in Canada: Initial results from the FH Canada national registry

Liam R. Brunham [a, *], Isabelle Ruel [b], Etienne Khoury [h], Robert A. Hegele [c], Patrick Couture [d], Jean Bergeron [d], Alexis Baass [e, f], Robert Dufour [g], Gordon A. Francis [a], Lubomira Cermakova [a], G.B. John Mancini [i], James M. Brophy [b, j], Dianne Brisson [h], Daniel Gaudet [h], Jacques Genest [b, j, **]

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3185 patients in the database -2018:

- 3108 HeFH
- 14 HoFH
- 63 patients with other lipoprotein disorders (*ABCA1, SMPD1, APOAI, LCAT* mutations)

Table 1
Baseline characteristics of patients in the Canadian FH Registry.

Characteristic	HeFH	HoFH
N	3108	14
Age, years (mean ± SD) (n = 3022)	43 ± 17	38 ± 15
DLCNC[a] score (mean ± SD) (n = 3108)	5.7 ± 5.2	15.2 ± 5.2
Male sex (%) (n = 3097)	52.5%	57.1%
BMI[b], kg/m^2 (mean ± SD) (n = 2912)	26.0 ± 5.0	26.1 ± 4.0
Coronary artery disease (%) (n = 1857)	16.6%	57.1%
Systemic hypertension (%) (n = 2480)	21.1%	28.6%
Type 2 diabetes (%) (n = 1758)	5.6%	0%
Current smoker (%) (n = 2360)	17.0%	12.5%
Total cholesterol, mmol/L (mean ± SD) (n = 3043)	8.09 ± 1.83	13.0 ± 5.13
LDL-C, mmol/L (mean ± SD) (n = 2992)	6.06 ± 1.74	11.2 ± 5.35
HDL-C, mmol/L (mean ± SD) (n = 3037)	1.21 ± 0.37	1.03 ± 0.27
Triglycerides, mmol/L (median [interquartile range]) (n = 3035)	1.60 [1.03–2.30]	1.03 [0.85–2.6]
Apolipoprotein B, g/L (mean ± SD) (n = 1419)	1.48 ± 0.37	2.55 ± 0.83
Lipoprotein(a), mg/L (median [interquartile range]) (n = 994)	263 [81.0–678.0]	326 [97.7–1220.0]
Lipid-lowering therapy[c] (%) (n = 2293)	59.1%	78.6%
Any statin (%) (n = 2293)	51.4%	71.4%
High intensity statin (%) (n = 2293)	9.9%	57.1%

The number in parenthesis for each row indicates the number of HeFH participants for whom the data field was captured.
HeFH = heterozygous familial hypercholesterolemia. HoFH = homozygous familial hypercholesterolemia. Lipid levels were at the time of entry to registry.

[a] Dutch Lipid Clinic Network Criteria.
[b] Body mass index.
[c] Use of lipid lowering therapy is at time of entry into the Registry.

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2- Meta-analysis of FH prevalence

BMJ Open

Estimating the prevalence of heterozygous familial hypercholesterolaemia: a systematic review and meta-analysis

Leo E Akioyamen,[1,2] Jacques Genest,[3,4] Shubham D Shan,[1,2] Rachel L Reel,[1] Jordan M Albaum,[1] Anna Chu,[1,2] Jack V Tu[1,2,5]

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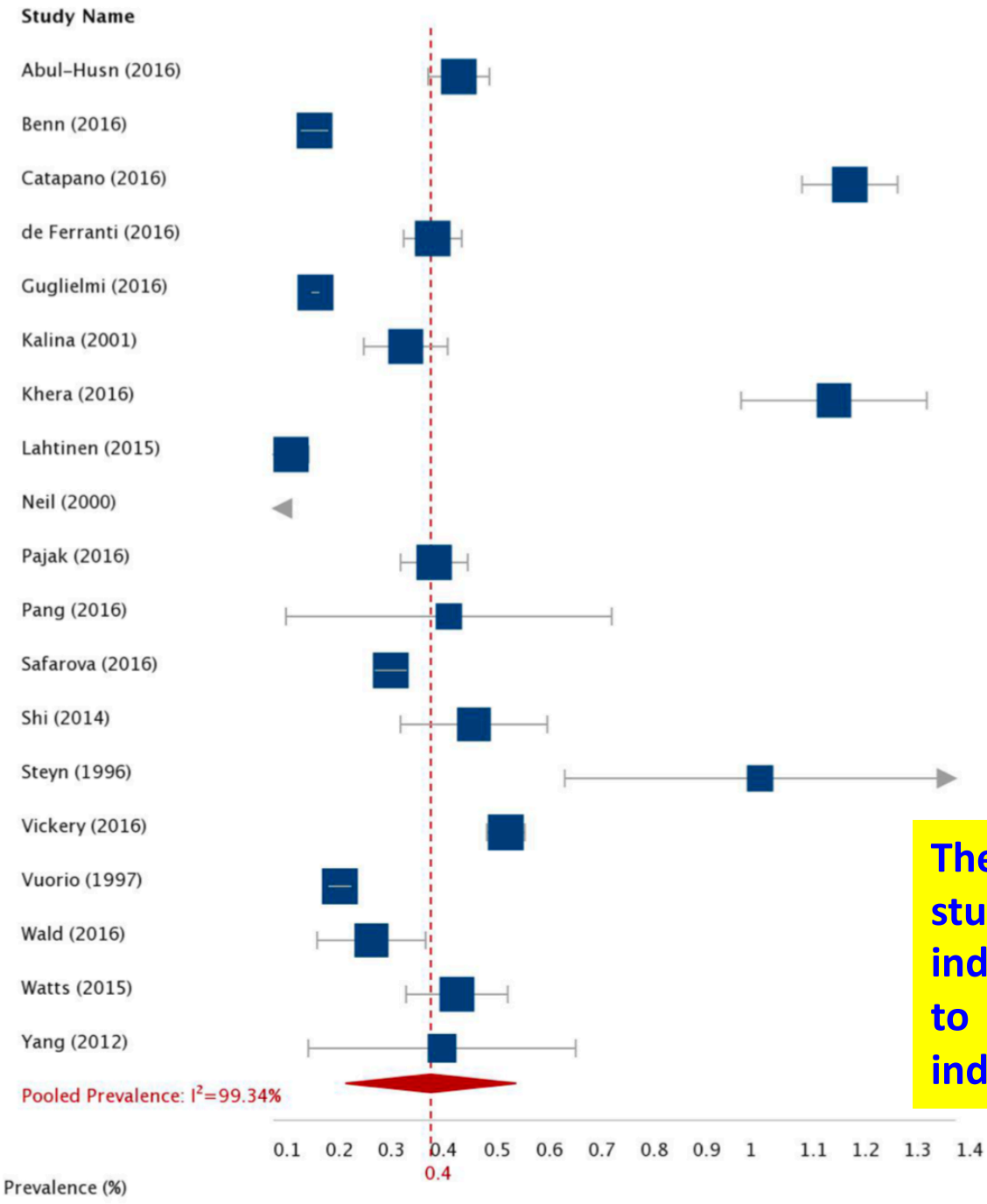

The pooled prevalence of FH from 19 studies including 2 458 456 unique individuals was 0.40% (95% CI 0.29% to 0.52%) = frequency of 1 in 250 individuals.

Akioyamen LE et al. BMJ Open. 2017;7:e0164615.

3- New Canadian FH definition

Canadian Journal of Cardiology 34 (2018) 1210–1214

Training/Practice

Contemporary Issues in Cardiology Practice

Simplified Canadian Definition for Familial Hypercholesterolemia

Isabelle Ruel, PhD,[a] Diane Brisson, PhD,[b] Sumayah Aljenedil, MD,[a] Zuhier Awan, MD, PhD,[c] Alexis Baass, MD, MSc,[d,e] Alexandre Bélanger, BSc,[a] Jean Bergeron, MD, MSc,[f] David Bewick, MD,[g] James M. Brophy, MD, PhD,[a,h] Liam R. Brunham, MD, PhD,[i] Patrick Couture, MD, PhD,[f] Robert Dufour, MD, MSc,[j] Gordon A. Francis, MD,[i] Jiri Frohlich, MD,[k] Claude Gagné, MD,[f] Daniel Gaudet, MD, PhD,[b] Jean C. Grégoire, MD,[l] Milan Gupta, MD,[m] Robert A. Hegele, MD,[n] G.B. John Mancini, MD,[o] Brian W. McCrindle, MD,[p] Jing Pang, PhD,[q] Paolo Raggi, MD, PhD,[r] Jack V. Tu, MD, PhD,[s] Gerald F. Watts, DSc, MD,[q,t] and Jacques Genest, MD[a,h]

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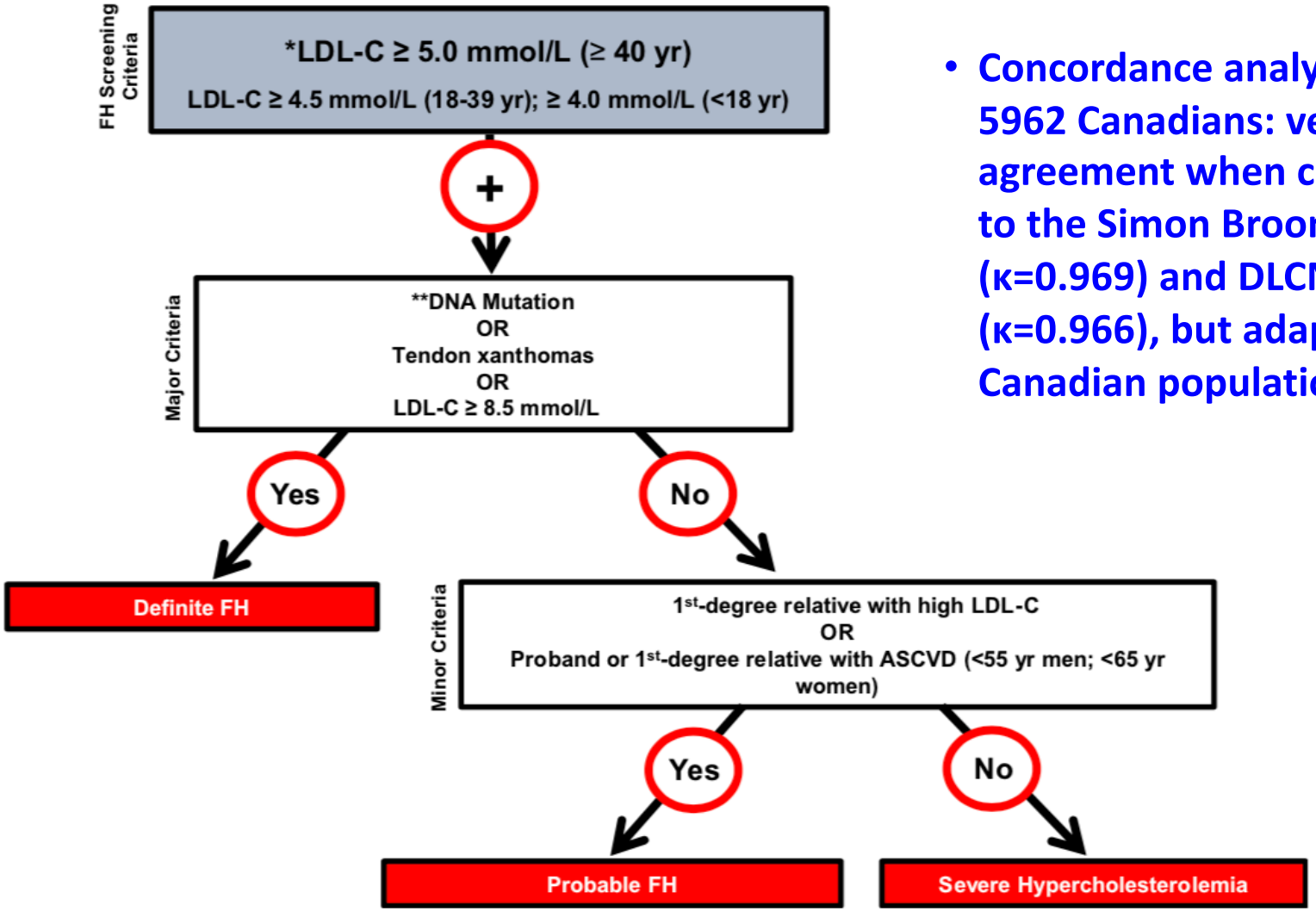
3- New Canadian FH definition



- Concordance analyses in 5962 Canadians: very good agreement when compared to the Simon Broome (κ=0.969) and DLCN (κ=0.966), but adapted to the Canadian population

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4- Algorithm to impute the baseline LDL-C when patient is on treatment and baseline LDL-C is unknown

When baseline LDL-C values are unknown, the database has an algorithm that can impute a LDL-C value from the LDL-C on treatment:

Table 1. Expected percent reduction in LDL-C according to dose and statin and ezetimibe.[a]

Medication	Mean reduction by dose: percent change from baseline (divide LDL-C by this factor)				
	5 mg	10 mg	20 mg	40 mg	80 mg
Rosuvastatin	−40 (0.60)	−46 (0.54)	−52 (0.48)	−55 (0.45)	–
Atorvastatin	–	−37 (0.63)	−43 (0.57)	−48 (0.52)	−51 (0.49)
Simvastatin	−26 (0.74)	−30 (0.70)	−38 (0.62)	−41 (0.59)	−47 (0.53)
Lovastatin	–	−21 (0.79)	−27 (0.73)	−31 (0.69)	−40 (0.60)
Pravastatin	–	−20 (0.80)	−24 (0.76)	−30 (0.70)	−36 (0.64)
Fluvastatin	–	–	−22 (0.78)	−25 (0.75)	−35 (0.65)
Ezetimibe alone	–	−20 (0.80)	–	–	–
Ezetimibe 10 mg added to a statin	−20 (0.80)	−20 (0.80)	−20 (0.80)	−20 (0.80)	−20 (0.80)

[a] Data derived from Hou et al. *(30)*.

PCSK9 inhibitors: Approx. 60 % decrease in LDL-C on any statin +/- Ezetimibe treatment (divide LDL-C by 0.4)**

Ruel I *et al.* Imputation of baseline LDL-C concentration in patients with FH on statins or ezetimibe. *Clinical Chemistry*. 2018;64(2):355-362

5- FH Canada “App” - Apple and Android

CardioRisk Calculator

By The University of British Columbia

This app is only available on the App Store for iOS devices.

View More by This Developer

http://www.circl.ubc.ca

or free download from app stores

Description

CardioRisk Calculator™ simplifies cardiovascular risk stratificatio
application.

CardioRisk Calculator Support ›

What's New in Version 1.3.3

–Addition of Bruits/AAA/Pulse Deficit fields
–Updated Dyslipidemia Risk Calculator
–Updated FH Calculator

...More

This app is designed for both iPhone and iPad

Free

Category: Medical
Updated: Dec 17, 2017
Version: 1.3.3
Size: 2.1 MB
Language: English
Seller: The University of British Columbia – Okanagan
© 2017 The University of British Columbia
You must be at least 17 years old to download this app.
Frequent/Intense Medical/Treatment Information

Compatibility: Requires iOS 7.1 or later. Compatible with iPhone, iPad, and iPod touch.

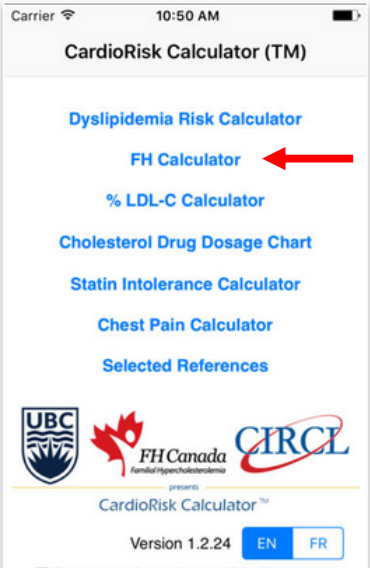
Customer Ratings

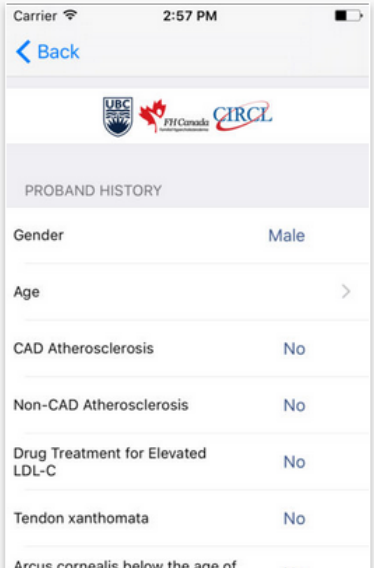
This application hasn't received enough ratings to display a summary.

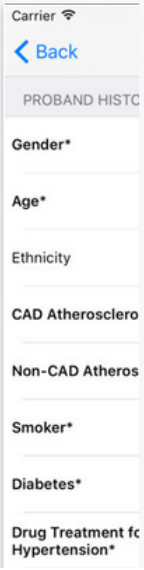
More by The University of British Columbia

Screenshots

iPhone | iPad







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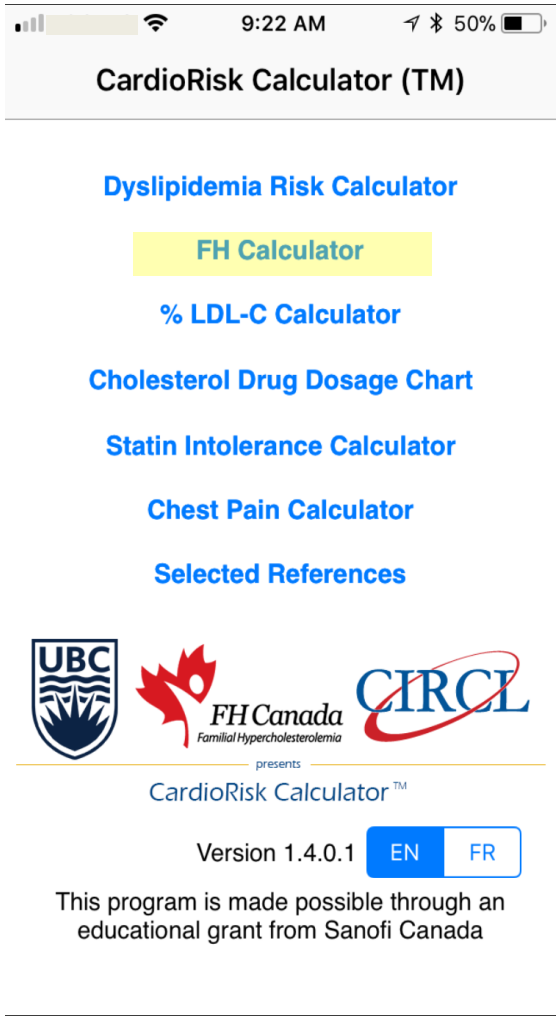
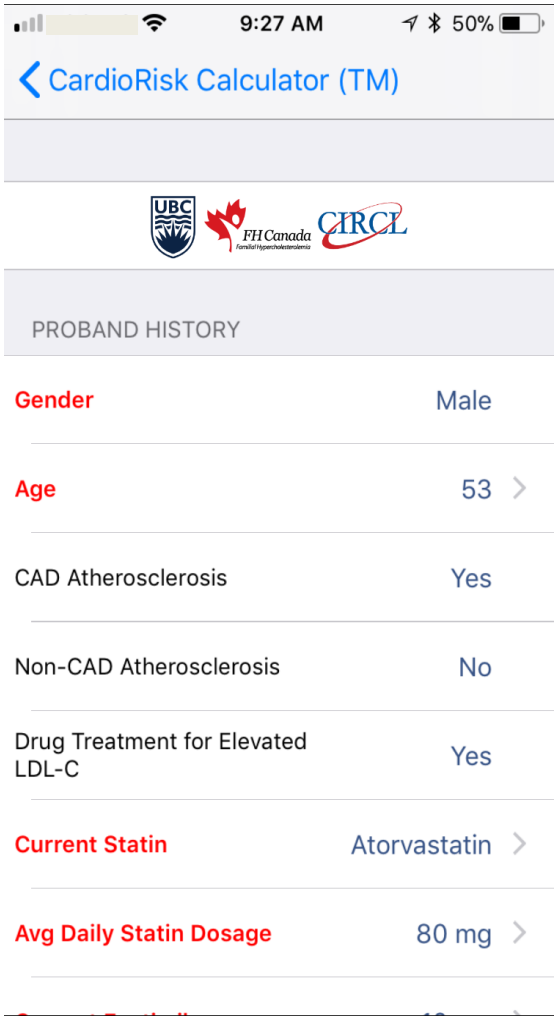
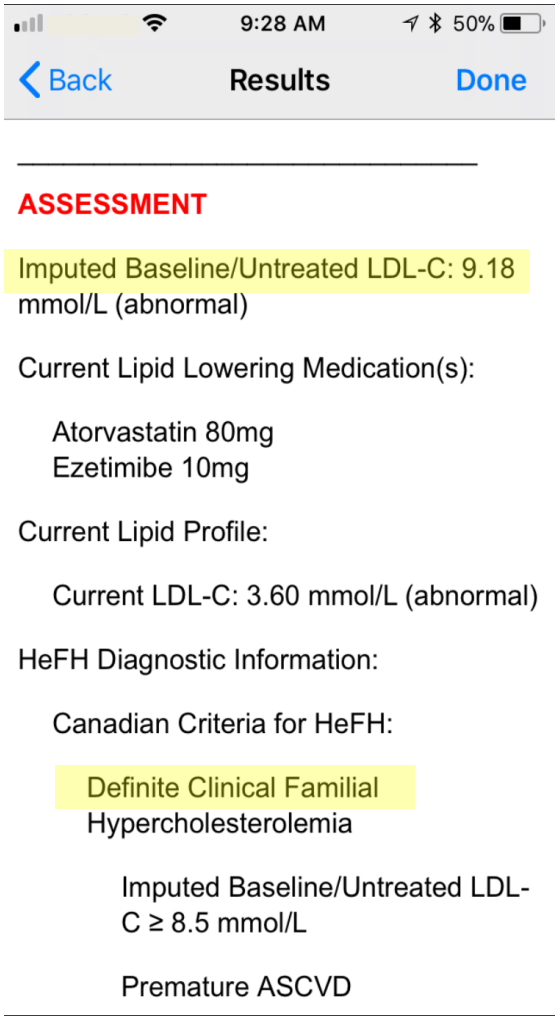
Tools to facilitate the diagnosis of FH







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6- Canadian Position Statement on FH

2018 Update of the Canadian Cardiovascular Society Position Statement on FH to be published in the *Canadian Journal of Cardiology* DEC 2018

Canadian Journal of Cardiology 34 (2018) 1553–1563

Society Position Statement

Canadian Cardiovascular Society Position Statement on Familial Hypercholesterolemia: Update 2018

Primary Panel: Liam R. Brunham, MD, PhD,[a,b] Isabelle Ruel, PhD,[c] Sumayah Aljenedil, MD,[c] Jean-Baptiste Rivière, PhD,[c] Alexis Baass, MD, MSc,[d,e] Jack V. Tu, MD, PhD,[f,†] G.B. John Mancini, MD,[a] Paolo Raggi, MD, PhD,[g] Milan Gupta, MD,[h] Patrick Couture, MD, PhD,[i] Glen J. Pearson, PharmD,[g] Jean Bergeron, MD, MSc,[i] Gordon A. Francis, MD,[a,j] Brian W. McCrindle, MD, MPH,[k] Katherine Morrison, MD,[l] Julie St-Pierre, MD, PhD,[m] Mélanie Henderson, MD, PhD,[n] Robert A. Hegele, MD, (Co-chair),[o] Jacques Genest, MD, (Co-chair),[c,d] **Secondary Panel:** Jeannette Goguen, MD,[p] Daniel Gaudet, MD, MSc,[q] Guillaume Paré, MD, MSc,[r] Jacques Romney, MD,[s] Thomas Ransom, MD, MSc,[t] Sophie Bernard, MD, PhD,[e,u] Pamela Katz, MD,[v] Tisha R. Joy, MD,[w] David Bewick, MD,[x] and James Brophy, MD, PhD[c,d]

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†Deceased.

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The disclosure information of the authors and reviewers is available from the CCS on their guidelines library at www.ccs.ca.

This statement was developed following a thorough consideration of medical literature and the best available evidence and clinical experience. It represents the consensus of a Canadian panel comprised of multidisciplinary experts on this topic with a mandate to formulate disease-specific recommendations. These recommendations are aimed to provide a reasonable and practical approach to care for specialists and allied health professionals obliged with the duty of bestowing optimal care to patients and families, and can be subject to change as scientific knowledge and technology advance and as practice patterns evolve. The statement is not intended to be a substitute for physicians using their individual judgement in managing clinical care in consultation with the patient, with appropriate regard to all the individual circumstances of the patient, diagnostic and treatment options available and available resources. Adherence to these recommendations will not necessarily produce successful outcomes in every case.

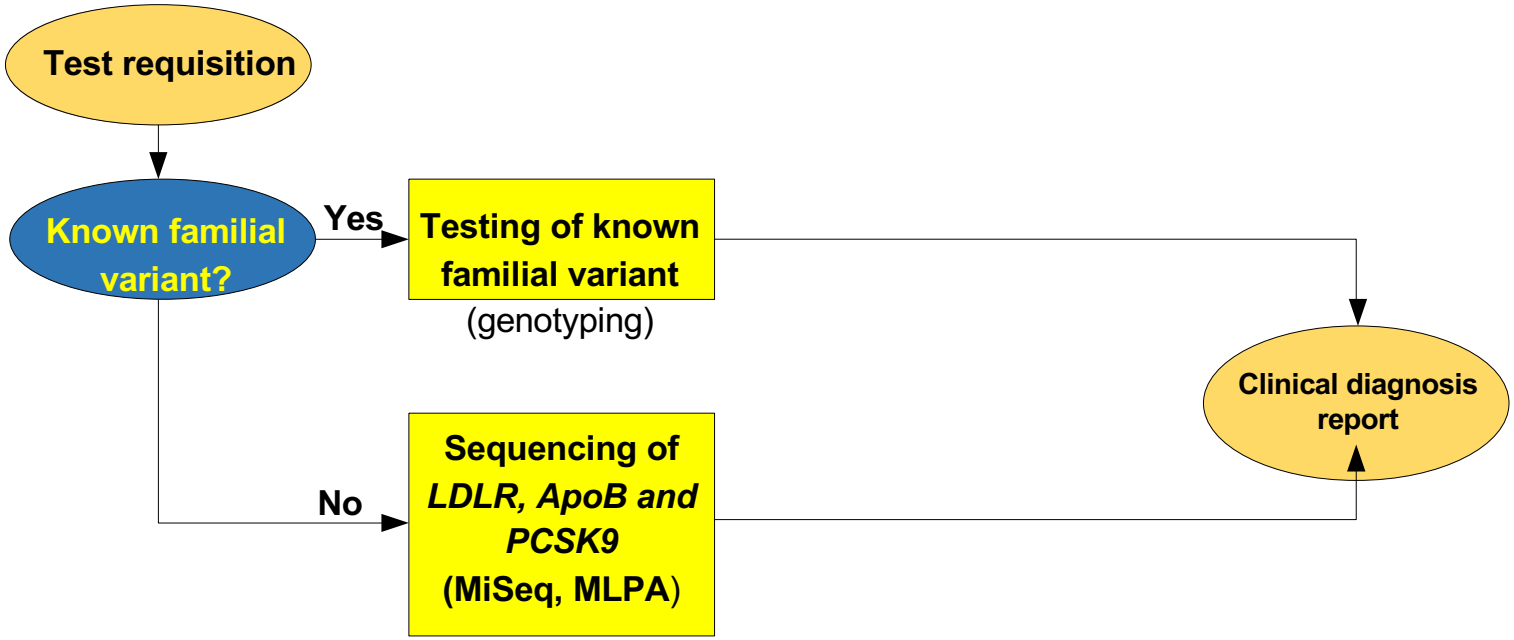
https://doi.org/10.1016/j.cjca.2018.09.005

HF Canada Hypercholestérolémie Familiale FH Canada Familial Hypercholesterolemia

7- Molecular Diagnosis of FH: complete DNA sequencing at MUHC

The McGill University Health Centre is the only CLIA-certified clinical molecular genetics lab in Canada

Clinical Laboratory Improvement Amendments - CLIA certification from FDA, CMS and CDC



HF Canada Hypercholestérolémie Familiale
FH Canada Familial Hypercholesterolemia

7- Molecular Diagnosis of FH: complete DNA sequencing at MUHC

OPTILAB Montréal-CUSM — Centre universitaire de santé McGill — McGill University Health Centre

Core Molecular Diagnostic Laboratory (CLIA #99D1042152)
1001 Decarie boul., E05.5051
Montreal, QC, H4A 3J1 Canada
Tel: 514-934-1934 x23383 / x23298 Fax: 514-843-1661

Patient Information:

Name (Last, First):

Birth date (YYYY-MM-DD): / /

Name of Referring Physician:

Physician's Specialty:

PATIENT STAMP OR LABEL HERE

Familial Hypercholesterolemia Panel – Testing Eligibility Criteria Form

Minimum criteria required for testing to be appropriate are listed below. Please complete and provide any relevant familial and clinical information. If the patient does not fulfil the criteria and you still feel that testing should be performed, please contact the laboratory or https://www.fhcanada.net to discuss testing of the sample.

Confirm diagnosis *(Indications and minimum criteria required for testing)*:

Untreated elevated LDL-cholesterol levels (not due to secondary causes).[1,2]

- ☐ Untreated LDL-cholesterol levels ≥ 5.0 mmol/L for age 40 yr and over – Specify level: mmol/L
- ☐ Untreated LDL-cholesterol levels ≥ 4.5 mmol/L for age between 18 yr and 39 yr – Specify level: mmol/L
- ☐ Untreated LDL-cholesterol levels ≥ 4.0 mmol/L for age under 18 yr – Specify level: mmol/L

AND at least one of the following:

Major Criteria (definite FH)

- ☐ Tendon xanthomas in proband.
- ☐ Known FH-causing DNA mutation in a first-degree relative.
- ☐ High LDL-cholesterol in proband (≥ 8.5 mmol/L).

Minor Criteria (probable FH)

- ☐ First-degree relative with high LDL-cholesterol (not due to secondary causes).[1]
- ☐ Proband or first-degree relative with early onset atherosclerotic cardiovascular disease (men under 55 yr; women under 65 yr).

[1]Secondary causes of high LDL-cholesterol should be ruled out (severe or untreated hypothyroidism, nephrotic syndrome, hepatic disease [primary biliary cirrhosis], or medication especially antiretroviral agents).
[2]If baseline LDL-cholesterol is unknown, an imputed level can be derived using the CardioRisk Calculator (http://www.circl.ubc.ca/cardiorisk-calculator.html).

OPTILAB Montréal-CUSM — Centre universitaire de santé McGill — McGill University Health Centre

PATIENT STAMP OR LABEL HERE

Molecular Genetics Requisition - CMDL

Core Molecular Diagnostic Laboratory (CLIA #99D1042152)
1001 Decarie boul., E05.5051
Montreal, QC, H4A 3J1 Canada
Tel: 514-934-1934 x23383 / x23298 Fax: 514-843-1661

Patient Information:

Name (Last, First):*

Birth date (YYYY-MM-DD):* / /

Gender:* ☐ Male ☐ Female ☐ Unknown

Father's name:

Mother's name:

For Canada only:

Provincial Health Card #:*

Issuing Province:*

Referring Physician:

Name (Last, First):*

License #:*

Institution:*

e-mail address:*

Address:*

Tel:* Fax:*

Genetic counsellor: *(Fax # to send results)*

Tel:* Fax:*

Signature:* Date:* / /

I acknowledge that the patient/guardian is aware of the benefits, limitations and risks associated with the requested test(s) and that I have obtained informed consent to perform genetic testing for this patient.
I authorize the laboratory to fax results to the number provided above.

Sample Information:

Collection date (YYYY-MM-DD):* / /

☒ Blood in EDTA (purple top tube): min 5 mL (2 mL for newborns)

☐ DNA: min 5 ug – Source:

☐ Amniotic fluid: min 10 mL

☐ Cultured amniocytes: 2 confluent T25 flasks

☐ Direct CVS: min 10 mg direct villi

☐ Cultured CVS: 2 confluent T25 flasks

☐ Tissue – Specify:

☐ Other – Specify:

CMDL - Laboratory use only:

Date - Time received: / /

h min

SAMPLE LABEL HERE

Sample type and # of tubes:

Patient #:

Test Requested ***(write below and check box(es) on page 2)***:*

FH panel

Reason for Testing:*

- ☐ Confirm diagnosis (symptomatic)
- ☐ Carrier testing (for recessive conditions)
- ☐ Predictive testing (for dominant conditions)
- ☐ Prenatal testing (maternal sample required)
- ☐ Other – Specify:

Reason for expedited testing *(if applicable)*:

- ☐ Pregnancy (Gestational age: weeks on / /)
- ☐ Other reason – Specify:

Familial Variant Analysis:

For cases where a familial variant is known, please complete below and attach a copy of the proband's report. If the familial variant was not previously tested at the CMDL, please provide a sample from a family member known to be positive for this variant (i.e. positive control).

Gene (HGNC symbol):

Variant(s) (HGVS nomenclature):

CMDL Family number:

Name of proband:

Relationship to proband:

Pedigree/Clinical Information:

Please draw or attached pedigree and provide all relevant information.

Ethnicity:*

Ordering Checklist:

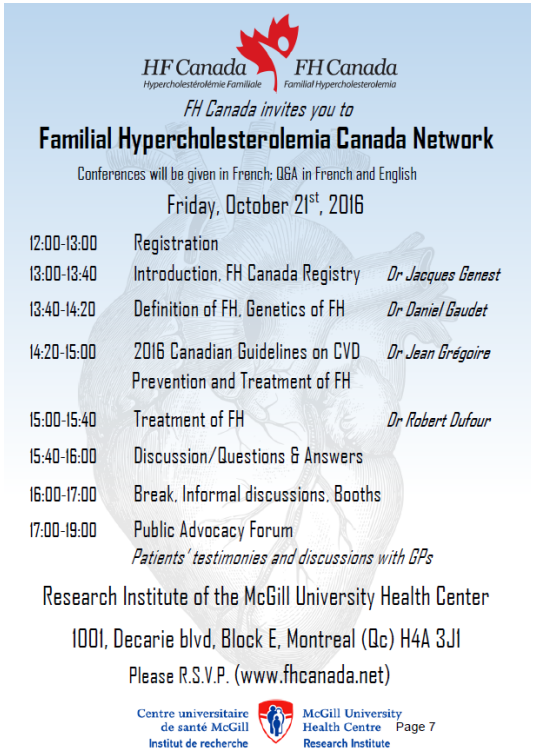
- ☐ Specimen tube labelled with at least two identifiers
- ☐ Completed test requisition (this form)
- ☐ Completed testing eligibility criteria form (if applicable)
- ☐ Consent form (or signature that consent form was obtained)

Required information. Samples will not be processed if information is missing.

HF Canada Hypercholestérolémie Familiale — FH Canada Familial Hypercholesterolemia

Other initiatives

- **FH Canada Network: annual accredited meetings, including a patient advocacy forum**



HF Canada Hypercholestérolémie Familiale — FH Canada Familial Hypercholesterolemia

FH Canada invites you to

Familial Hypercholesterolemia Canada Network

Conferences will be given in French; Q&A in French and English

Friday, October 21st, 2016

12:00-13:00	Registration	
13:00-13:40	Introduction, FH Canada Registry	*Dr Jacques Genest*
13:40-14:20	Definition of FH, Genetics of FH	*Dr Daniel Gaudet*
14:20-15:00	2016 Canadian Guidelines on CVD Prevention and Treatment of FH	*Dr Jean Grégoire*
15:00-15:40	Treatment of FH	*Dr Robert Dufour*
15:40-16:00	Discussion/Questions & Answers	
16:00-17:00	Break, Informal discussions, Booths	
17:00-19:00	Public Advocacy Forum	

Patients' testimonies and discussions with GPs

Research Institute of the McGill University Health Center

1001, Decarie blvd, Block E, Montreal (Qc) H4A 3J1

Please R.S.V.P. (www.fhcanada.net)

Centre universitaire de santé McGill Institut de recherche — McGill University Health Centre Research Institute

CCRN Canadian Collaborative Research Network — HF Canada Hypercholestérolémie Familiale — FH Canada Familial Hypercholesterolemia

Familial Hypercholesterolemia: How to Recognize and Manage Patients in Your Practice?

REGISTRATION

Early bird: $50.00

Valid until September 20 2017

$75 starting September 21

Registration closes: October 18 2017.

Please R.S.V.P. at www.ccrnmd.com

Friday, October 20, 2017

12:00-6:30 pm

St. Paul's Hospital,
1081 Burrard Street, Vancouver, BC, V6Z 1Y6
The Cullen Family Lecture Theatre, Room 1477,
Providence, Level 1

CHAIR

Liam R. Brunham, MD, PhD, FRCPC, FACP
Assistant Professor of Medicine, University of British Columbia
Scientist, UBC Centre for Heart Lung Innovation
Physician, Healthy Heart Program Prevention Clinic, St. Paul's Hospital
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Brampton, ON

Sanja Karalic, MSc, MD, CCFP
Clinical teaching instructor, Department of Family Medicine, University of British Columbia
Vancouver, BC

AGENDA

12:00 p.m.	**Registration / Lunch**	
1:00 p.m.	*Intro, Landscape of FH in Canada - role of FH registry*	Dr. Liam R. Brunham
1:20 p.m.	*Genetics of FH and role of genetic testing*	Dr. Jacques Genest
1:40 p.m.	*How to recognize and diagnose FH*	Dr. Gordon Francis
2:00 p.m.	**Q&A**	All faculty
2:20 p.m.	**Break**	
2:40 p.m.	*Treatment of FH including new and emerging therapies*	Dr. John Mancini
3:00 p.m.	*International Perspectives on FH*	Dr. Joshua Knowles
3:40 p.m.	**Discussion/Questions & Answers**	All speakers
4:00 p.m.	**Break, Informal discussions, Booths, Nutrition**	
4:30 p.m.	*Patient Forum: Patients' stories and discussions with medical doctors*	
6:30 p.m.	**Close**	

CCRN Canadian Collaborative Research Network

Managing FH Together: An Interchange Between Patients, Clinicians and Researchers

REGISTRATION

$75.00

Registration closes: October 18 2018.

REGISTER ONLINE

www.ccrnmd.com

Saturday, October 20th 2018

8:30 a.m. – 12:30 p.m.

Li Ka Shing Knowledge Institute
Allan Waters Family Auditorium
209 Victoria St. Toronto, ON

CO-CHAIRS

Liam R. Brunham
MD, PhD, FRCPC, FACP
Assistant Professor of Medicine, University of British Columbia
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"Learning Through Understanding"
Toronto, ON

Marla Shapiro
CM, CCFP, MHSC, FRCPC, FCFP, NCMP
Professor, DFCM
University of Toronto
Toronto, ON

Intended for family physicians and cardiovascular specialists

This session will review the newly revised Canadian Cardiovascular Society position statement on Familial Hypercholesterolemia (FH), including aspects of screening, diagnosis, and treatment. The role of registries for patients with FH will also be discussed.

AGENDA

7:45 a.m.	*Breakfast and registration*	
8:30 a.m.	Introduction	Liam Brunham
8:40 a.m.	The genetics of FH made simple	Robert Hegele
9:00 a.m.	The risk associated with FH	TBC
9:20 a.m.	Registries and cascade screening	Liam Brunham
9:40 a.m.	Panel discussion and audience participation	
10:00 a.m.	*Break*	
10:35 a.m.	Case Presentation	Omar Razek
10:45 a.m.	How to diagnose FH in your clinic?	Jacques Genest
11:05 a.m.	When to consider genetic testing?	TBC
11:25 a.m.	Evidence-based treatment of FH	Milan Gupta
11:45 a.m.	Panel discussion and audience participation	
12:05 p.m.	*Patient forum + boxed lunch*	

HF Canada Hypercholestérolémie Familiale — FH Canada Familial Hypercholesterolemia

Conclusion:
Opportunities and Challenges

- A better understanding of care gaps for patients with FH in Canada
- A simplified set of diagnostic criteria for the Canadian population with tools to aid in the diagnosis of FH.
- Access to molecular diagnosis
- Access to new medication

As the national registry further increases in size and scope, there will be opportunities to improve the diagnosis and care of patients with FH in Canada.

HF Canada
Hypercholestérolémie Familiale
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FH Canada registry is a unique network of more than 150 basic researchers, clinicians specializing in lipidology, endocrinology, pediatric endocrinology, obesity and cardiology, clinic coordinators and industry partners.

If you have any questions about the registry, you may visit our website at
www.fhcanada.net

You may also contact the **national coordinator**:
Isabelle Ruel PhD
Research Institute of the McGill University Health Centre
1001 Decarie blvd, Block E #E01.2123
Montreal, Quebec
H4A 3J1
514-934-1934, ext. 34852
info@fhcanada.net

McGill

Institut de recherche Centre universitaire de santé McGill
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If you have any questions about the registry, you may visit the website at *www.fhcanada.net*

Or e-mail Isabelle Ruel, info@fhcanada.net

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