HF Canada
Hypercholestérolémie Familiale

FH Canada
Familial Hypercholesterolemia

Canadian Familial Hypercholesterolemia Registry
Régistre Canadien d'hypercholestérolémie familiale

But du registre HF Canada

- **Le but du registre HF Canada est d'améliorer la detection et la prise en charge des patients et des membres de leur famille avec HF au Canada. Les patients avec d'autres désordres du métabolisme des lipoprotéines sont également inclus (initiative SMASH).**

- **Registre initié à l'Université de la Colombie-Britannique, et devenu national en 2014.**

- **Réseau de plus de 200 cliniciens et scientifiques, avec 19 centres académique au Canada.**

Clinicaltrials.gov: NCT02009345

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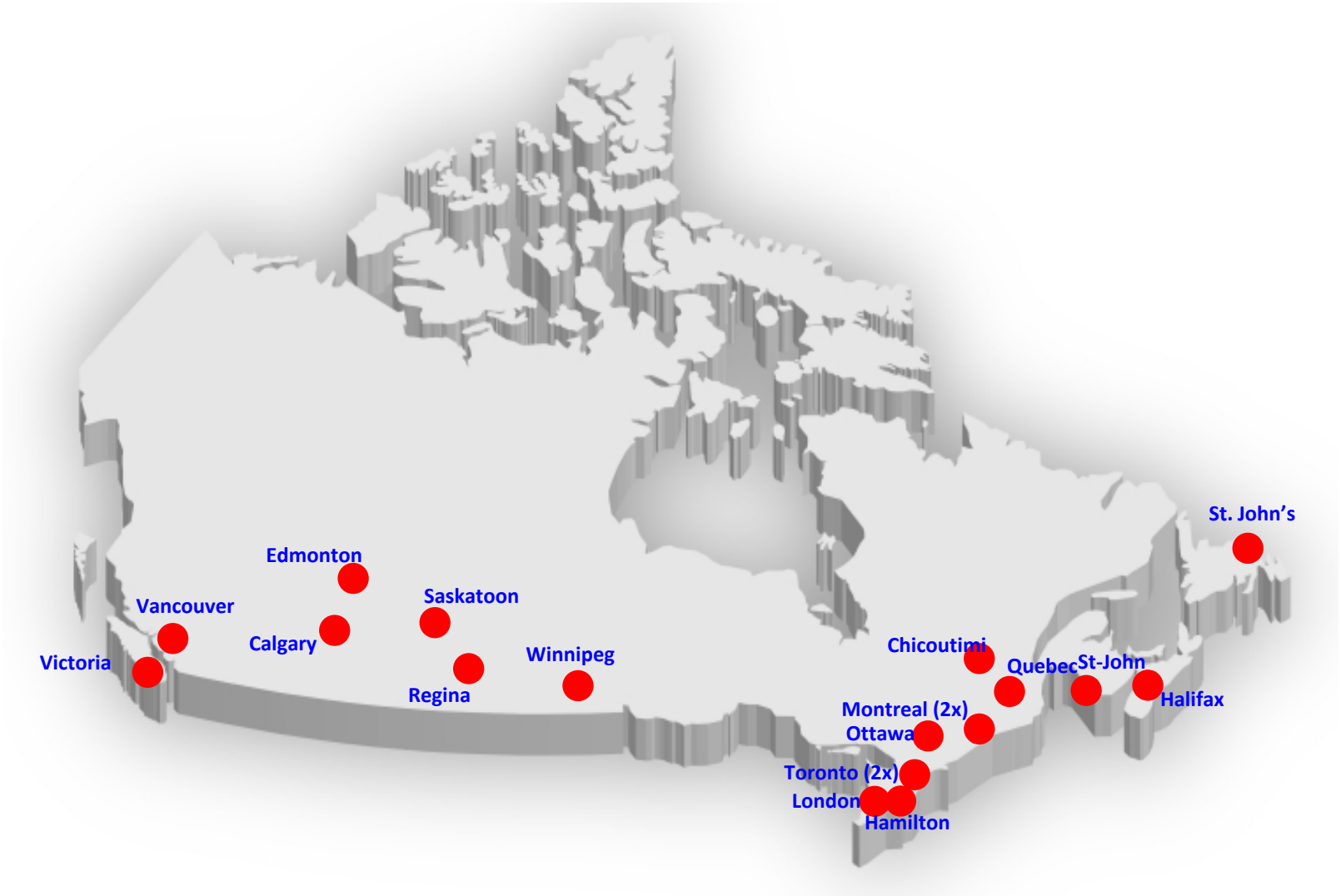
La MISSION du registre canadien d'HF est de rassembler une équipe multi-disciplinaire composée de médecins ainsi que de chercheurs fondamentaux et cliniciens afin d'améliorer les soins prodigués aux patients atteints de désordres lipidiques importants, et plus particulièrement d'HF, mais aussi de favoriser la collaboration en recherche.

Notre VISION est de créer un réseau de cliniques universitaires regroupant des spécialistes dans le domaine des lipides, des endocrinologues et des cardiologues afin d'offrir les meilleurs soins possibles aux patients et de favoriser la collaboration en recherche. Se basant sur le modèle de «réseau en étoile», le registre canadien d'HF s'étendra au sein de plusieurs communautés afin de rallier les médecins prodiguant les soins de santé primaires aux cliniques universitaires provinciales.

Nos OBJECTIFS sont d'améliorer les soins prodigués aux patients souffrant d'HF et de diminuer les maladies cardiovasculaires chez ces patients à haut risque.

Registre HF Canada: modèle “hub and spoke”



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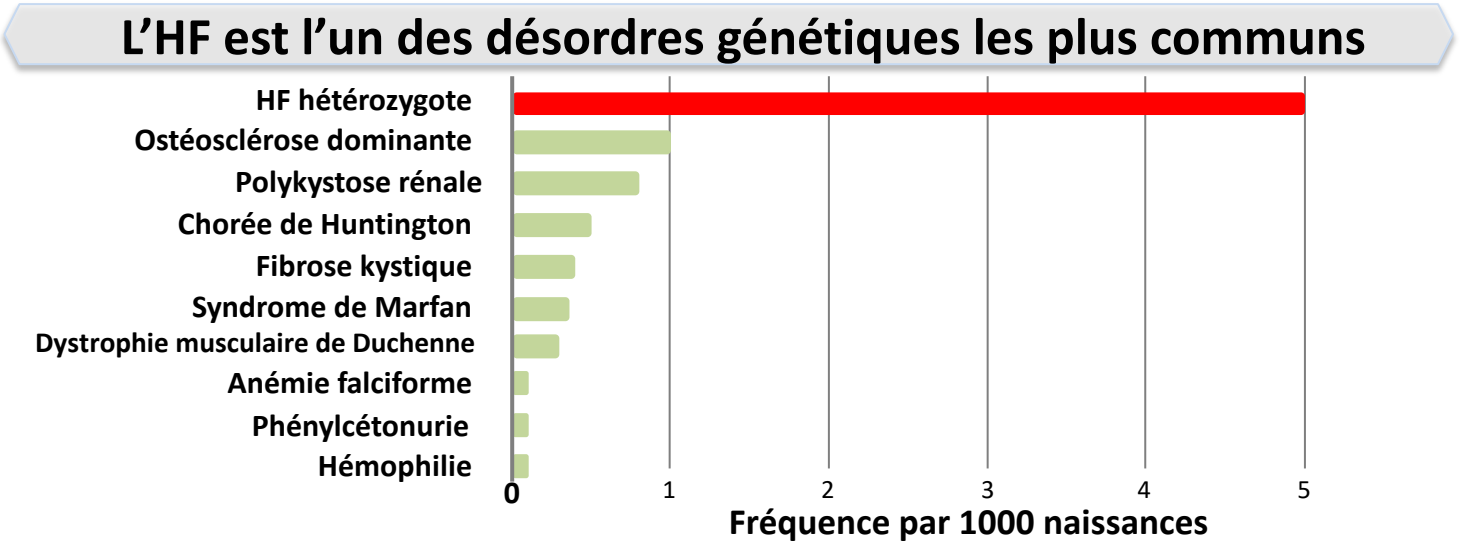
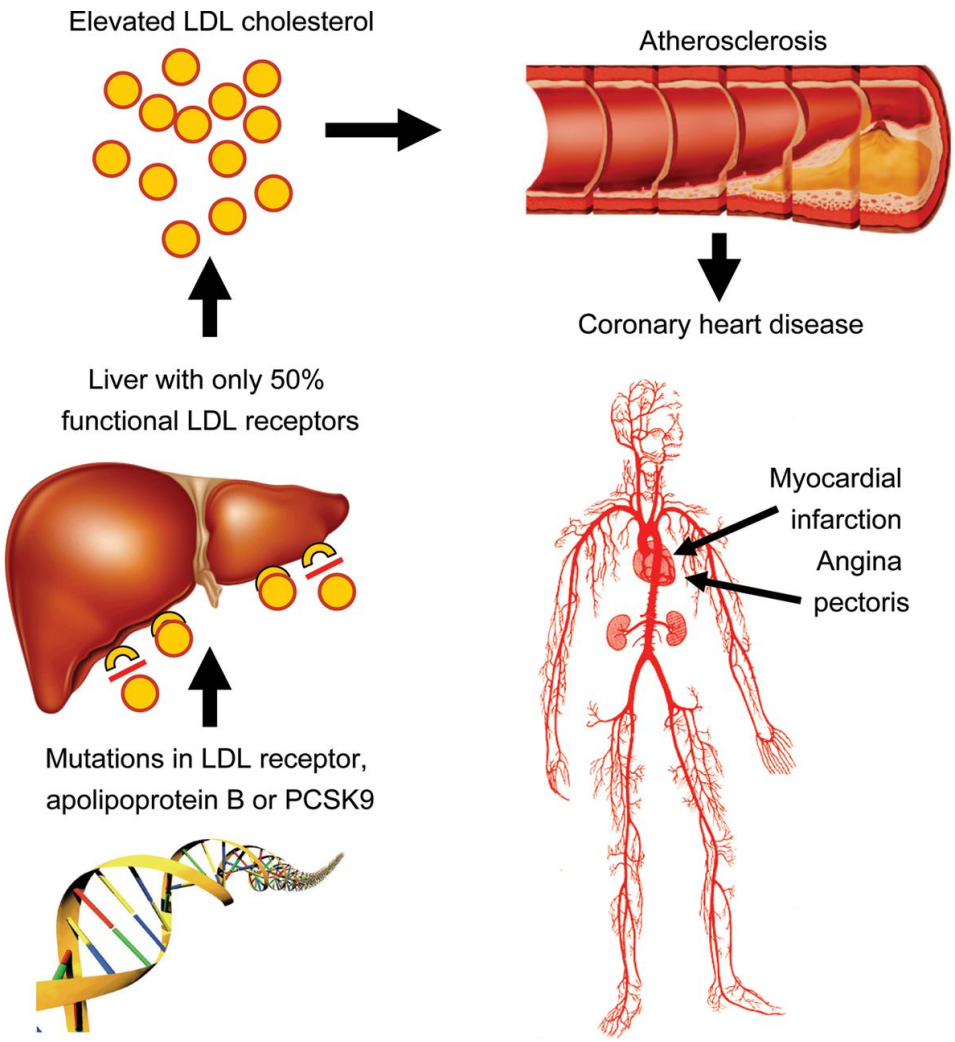


Nordestgaard B G et al. Eur Heart J 2013;34:3478-3490

- **Génétique, désordre autosomique co-dominant[1]**
- **Généralement due à des mutations dans le gène du récepteur des LDL[2,3]**
  - **> 1800 mutations**
  - **Prévalence: 1 sur 250 individus**
  - **Au moins 143,000 patients au Canada, avec moins de 10% des patients diagnostiqués**

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.

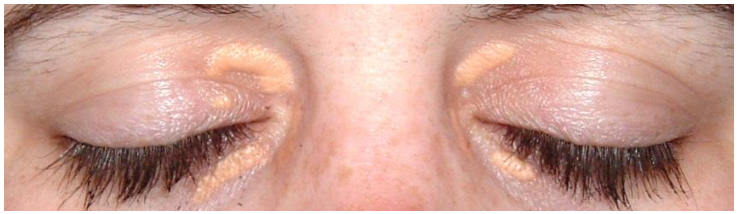
Pathophysiologie de l’ HeHF



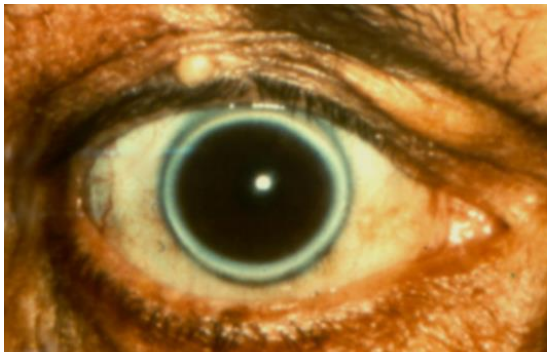
Nordestgaard B G et al. Eur Heart J 2013;34:3478-3490

European Heart Journal

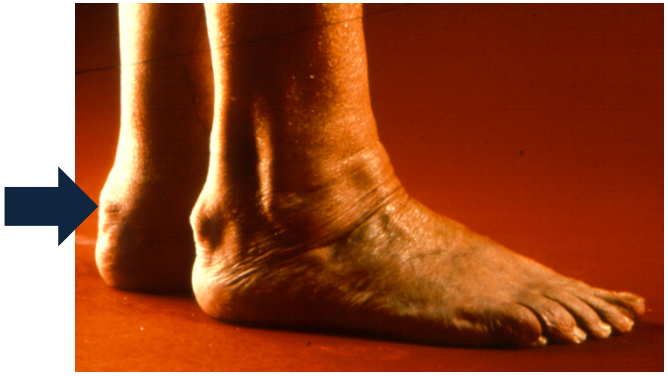
Manifestations cliniques



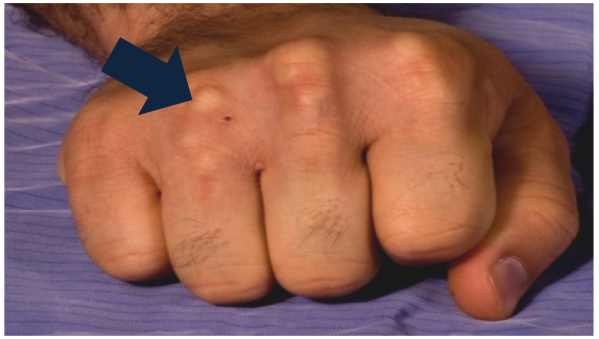
Xanthelasma bilatéraux



Arc cornéen



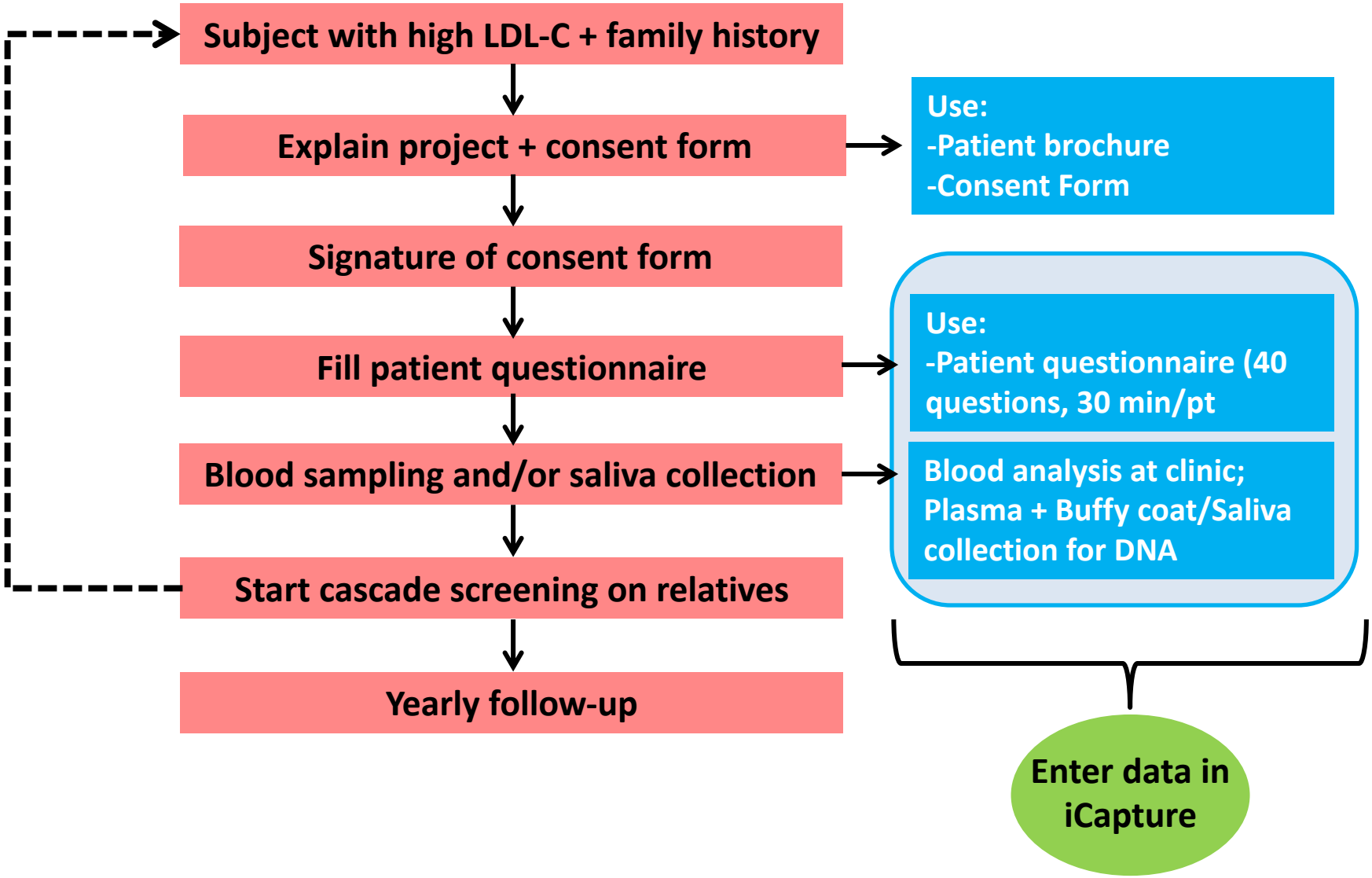
Xanthomes dans les tendons d'Achille



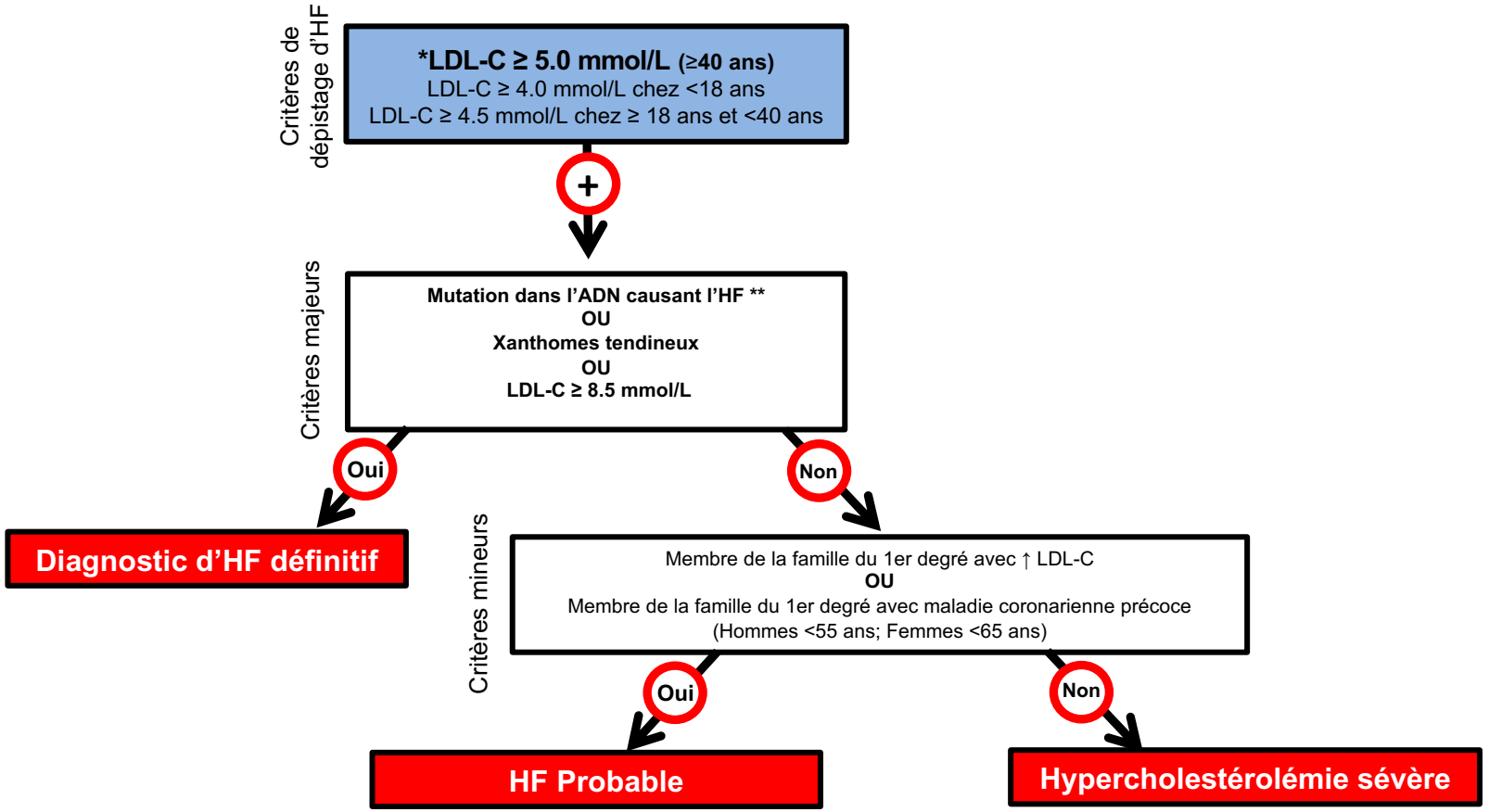
Xanthomes au niveau des tendons de l'extenseur des doigts

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.

En résumé, le registre c'est:



Critère d'inclusion:



*** Causes secondaires éliminées (syndrome néphrotique, ictère par obstruction, hypothyroïdie, médication, autres).**
**Mutations dans les gènes *LDLR*, *ApoB* ou *PCSK9*; La présence d'une mutation d'ADN chez le sujet-index ou chez un membre de la famille au premier degré est suffisante pour un diagnostique définitif d'HF. La décision de traiter ou non reste à la discrétion du médecin traitant.

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Données collectées dans le questionnaire au patient

Section 1: Données du patient

- Données démographiques (nom, adresse, date de naissance, sexe, appartenance ethnique déclarée, formulaire de consentement signé + date, méthode de contact préférée)
- Coordonnées du médecin de famille

Section 2: Examen médical antérieur

- Antécédents familiaux (antécédents familiaux de MCV et de taux élevé de LDL-C chez des parents au premier degré)
- Statut de fumeur (non fumeur, ex- ou fumeur, nb années, nb cigarettes / jour)
- Antécédents médicaux (hypertension, DM, coronaropathie, angine, etc.)
- Antécédents chirurgicaux (PCI, CABG, revascularisation artérielle)

Section 3: Examen physique

- Mesures standard (poids, taille, pression artérielle)
- Signes physiques d'HF (arc de la cornée <45 ans, xanthélasma, xanthomes)

Section 4: Données sur les médicaments

- Médicament hypolipidémiant (statine, ézétimibe, etc. avec dose et fréquence d'administration, intolérance aux statines)
- Médicament non hypolipidémiant (sur ordonnance ou en vente libre avec dose et fréquence)

Section 5: Données de laboratoire

- Date d'analyse
- Jeûne, oui / non
- Niveau de LDL-C sans traitement connu ou autodéclaré
- Glucose, hémoglobine A1C, cholestérol total, LDL-C, HDL-C, TG, CK, créatinine, AST, ALT, Lp(a), apoB

Section 6: Génétique

- Mutation(s) de l'ADN si connue (ADN isolé oui / non, gène + nom de la mutation)
- SNP score si connu

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Entrée de données dans iCapture

- Le Centre de recherche James Hogg situé à l'hôpital St-Paul de l'Université de la Colombie-Britannique à Vancouver fournit la **plate-forme iCAPTURE** qui est utilisée pour capturer les données du Registre HF Canada.

- La base de données utilise un serveur Oracle, est protégée par un pare-feu et gérée dans un réseau non public distinct. Elle est conforme aux normes FDA, Santé Canada, PHIA et PIPEDA. Tous les accès « utilisateur » sont enregistrés.

- Un identifiant unique est attribué au hasard à chaque patient (0 à 999999) et seulement cet identifiant est utilisé dans le registre national dé-identifié.

- Toutes les données anonymes du registre seront mises à disposition pour les statistiques sur l'HF au Canada, pour les études den économie de la santé et de résultats pour la santé, ce qui facilitera l'allocation des ressources et le contrôle de la qualité.

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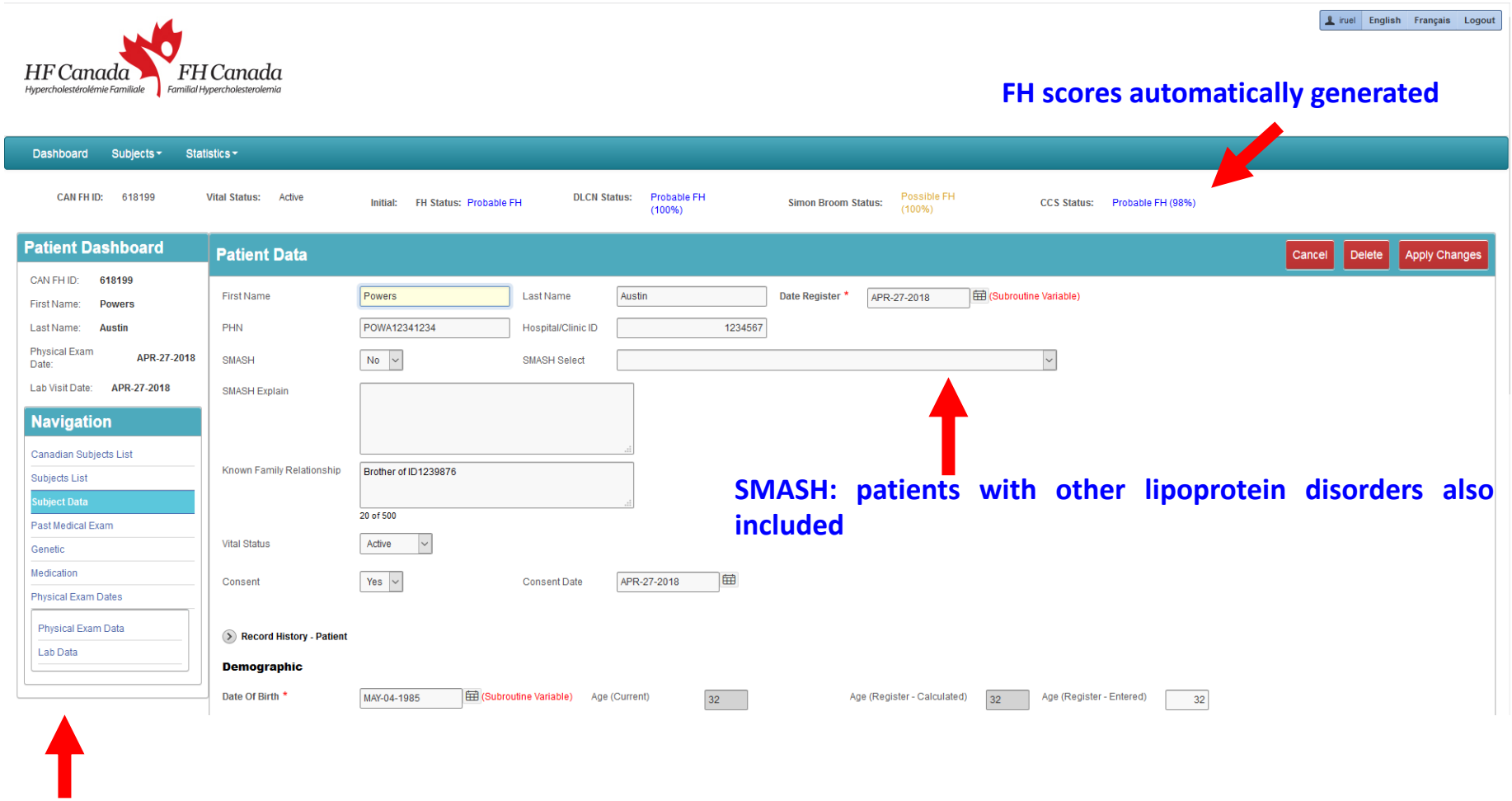
Base de données iCapture



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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La base de données comporte des algorithmes intégrés permettant de générer un score pour les critères FH les plus courants (Simon-Broome, Dutch Lipid Clinic Network (DLCN), définition canadienne).

Critère Simon Broome pour le diagnostic clinique de l'hypercholestérolémie familiale (HF)

Description			Critère
Présence d'une mutation génétique causant l'HF (gènes *LDLR*, *APOB*, *PCSK9*)			**Définitif**
LDL-C > 4.9 mmol/L chez l'adulte (> 4.0 mmol/L chez l'enfant de moins de 16 ans) **ou** Cholestérol total > 7.5 mmol/L chez l'adulte (> 6.7 mmol/L chez l'enfant de moins de 16 ans)	+	Xanthomes tendineux ou évidence de ces signes chez un apparenté de 1er ou 2eme degré.	**Définitif**
LDL-C > 4.9 mmol/L chez l'adulte (> 4.0 mmol/L chez l'enfant de moins de 16 ans) **ou** Cholestérol total > 7.5 mmol/L chez l'adulte (> 6.7 mmol/L chez l'enfant de moins de 16 ans)	+	Histoire familiale d'infarctus du myocarde avant l'âge de 50 ans pour un apparenté au 2eme degré ou avant l'âge de 60 ans pour un apparenté au 1er degré **ou** Histoire familiale de niveaux élevés de cholesterol total > 7.5 mmol/L chez un apparenté de 1er ou 2eme degré ou > 6.7 mmol/L chez un enfant de moins de 16 ans	**Possible**

Adapté de *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.*

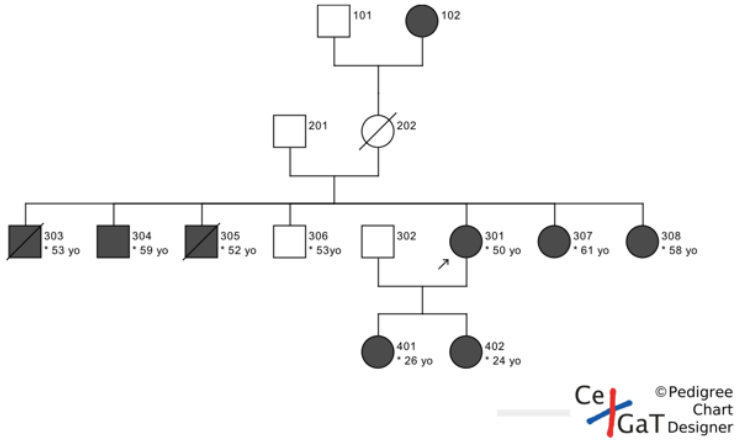
Critère Dutch Lipid Clinic Network pour le diagnostic clinique d'hypercholestérolémie familiale (HF)	
Groupe 1: Histoire familiale	
• Apparenté de 1er degré avec MCV prématurée (avant 55 ans chez l'homme, 60 ans chez la femme) **ou** • Apparenté de 1er degré avec LDL-C > 95th percentile	**1 point**
• Apparenté de 1er degré avec des xanthomes tendineux et/ou des arcs cornéens **ou** • Enfant de moins de 18 ans avec LDL-C > 95th percentile	**2 points**
Groupe 2: Histoire clinique	
• Patient avec maladie coronarienne prématurée (avant 55 ans chez l'homme, 60 ans chez la femme)	**2 points**
• Patient avec maladie vasculaire cérébrale ou périphérique (avant 55 ans chez l'homme, 60 ans chez la femme)	**1 point**
Groupe 3: Examen physique	
• Xanthomes tendineux	**6 points**
• Arcus cornéens présents avant 45 ans	**4 points**
Groupe 4: Analyses de laboratoire	
• 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**
Groupe 5: Analyse de l'ADN	
• Mutation génétique fonctionnelle connue pour être responsable de l'HF	**8 points**
DIAGNOSTIC d'HF	
• **Définitif** • **Probable** • **Possible** ***Le score le plus élevé par groupe doit être appliqué***	**9 points ou plus** **6-8 points** **3-5 points**

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

Sensibiliser la population sur l'HF: stratégie de dépistage en cascade

1- Les premiers patients à être recrutés sont ceux avec un taux élevé de LDL-C déjà suivi au site clinique.

2- Ensuite , les membres de la famille et d'autres patients non diagnostiqués (ex: les frères, sœurs et cousins) sont recrutés grâce au dépistage en cascade et sont dirigés au site HF Canada le plus proche (www.fhcanada.net).



3- Les nouveaux patients sont recrutés avec l'aide du site HF Canada et grâce à la sensibilisation de la population à l'HF.

Résultats récents:

1- Publication des résultats préliminaires du registre HF Canada

2- Caractérisation de la prévalence de l'HF (méta-analyse)

3- Validation d'une définition plus simple de l'HF pour les Canadiens

4- Validation d'un algorithme pour imputer le LDL-C de base lorsque le patient est sous traitement et que le LDL-C de référence est inconnu

5- Création d'une nouvelle «application» de FH Canada - Apple et Android, pour faciliter le diagnostic de l'HF

6- Mise à jour de l'énoncé de position de la SCC sur l'HF

7- Mise en place d'un test génétique d'HF - séquençage complet de l'ADN au CUSM.

1- Résultats préliminaires du registre HF Canada

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, **]

HF Canada Hypercholestérolémie Familiale

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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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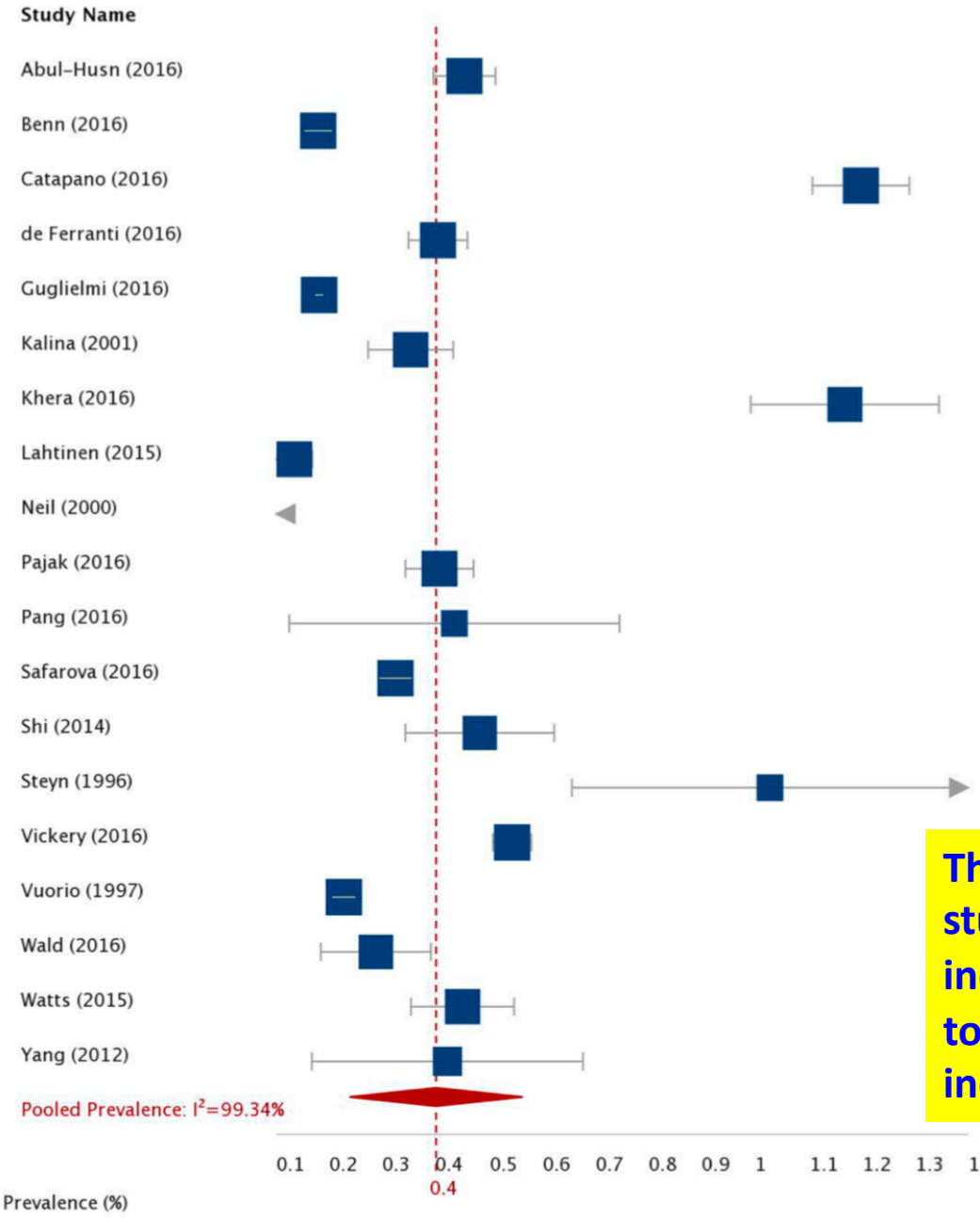
2- Caractérisation de la prévalence de l'HF (méta-analyse)

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]



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- Validation d'une définition plus simple de l'HF pour les Canadiens

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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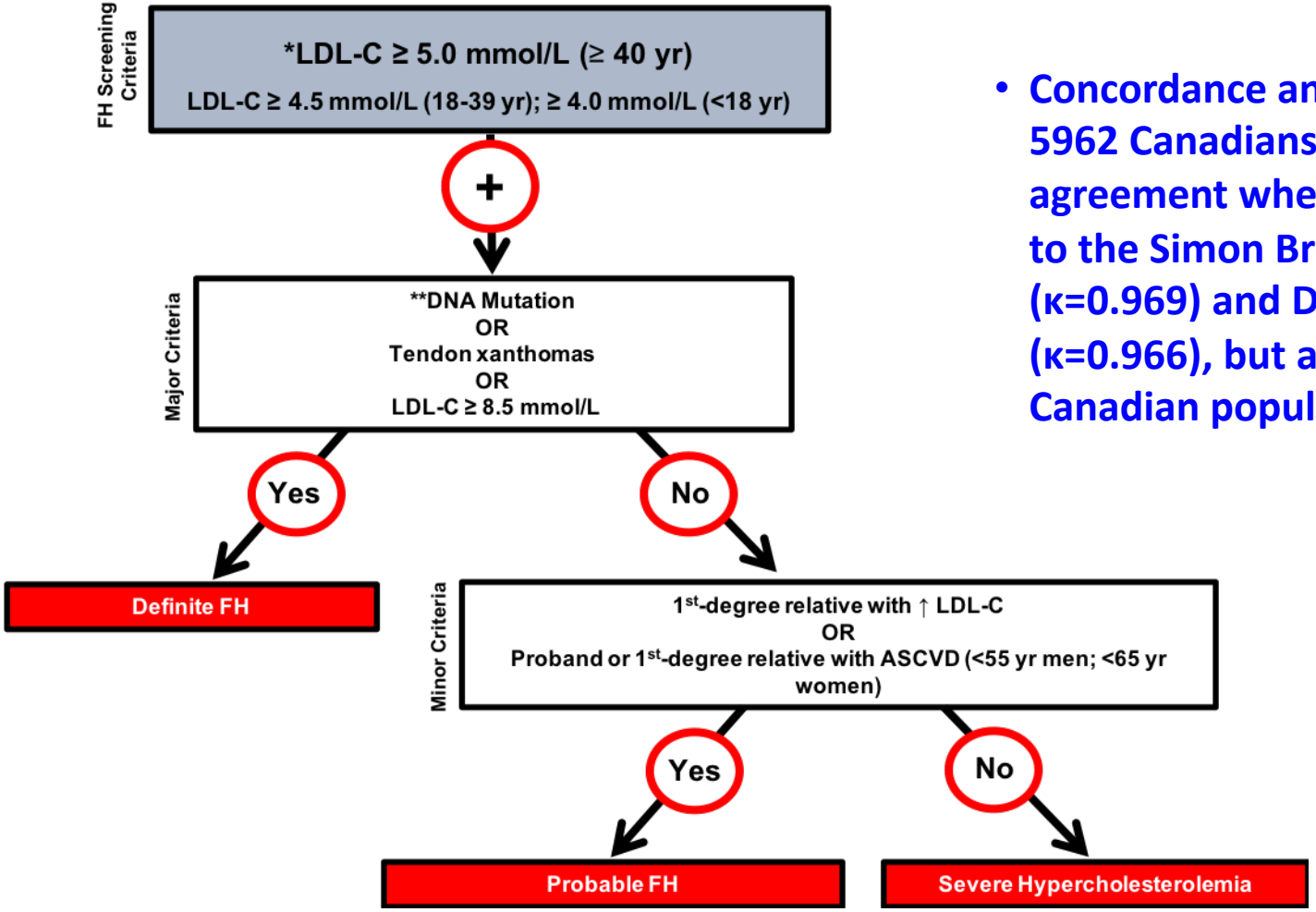

- **Concordance analyses in 5962 Canadians: very good agreement when compared to the Simon Broome (κ=0.969) and DLCN (κ=0.966), but adapted to the Canadian population**

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4- Validation d'un algorithme pour imputer le LDL-C de base

Lorsque les valeurs de référence de LDL-C ne sont pas connues, la base de données contient un algorithme qui permet d'attribuer une valeur de LDL- C non-traitée à partir de la valeur de LDL-C sous traitement:

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- Création d'une nouvelle «application» de FH Canada - Apple et Android, pour faciliter le diagnostic de l'HF

http://www.circl.ubc.ca

or free download from app stores

CardioRisk Calculator

View More by This Developer

By The University of British Columbia

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

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.

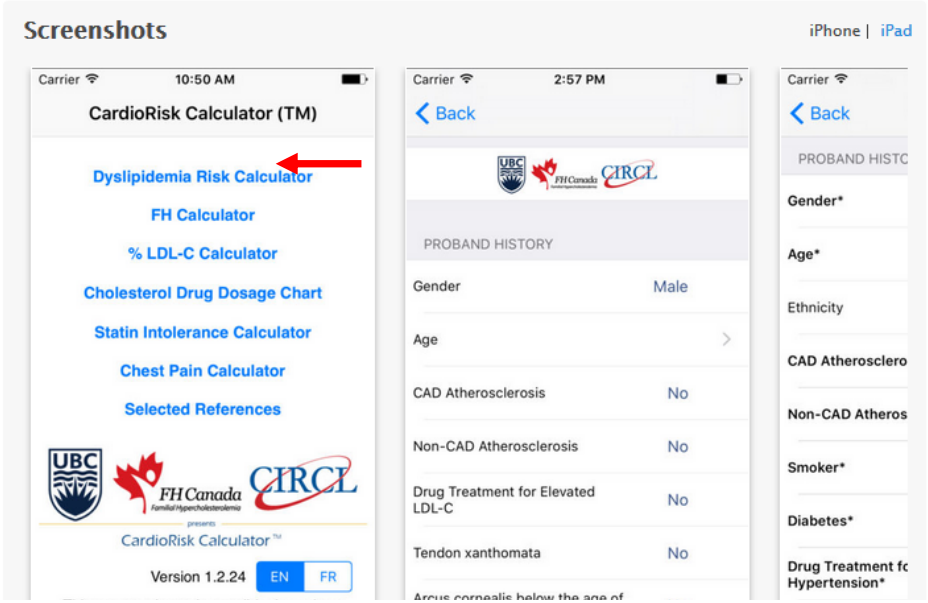
Customer Ratings

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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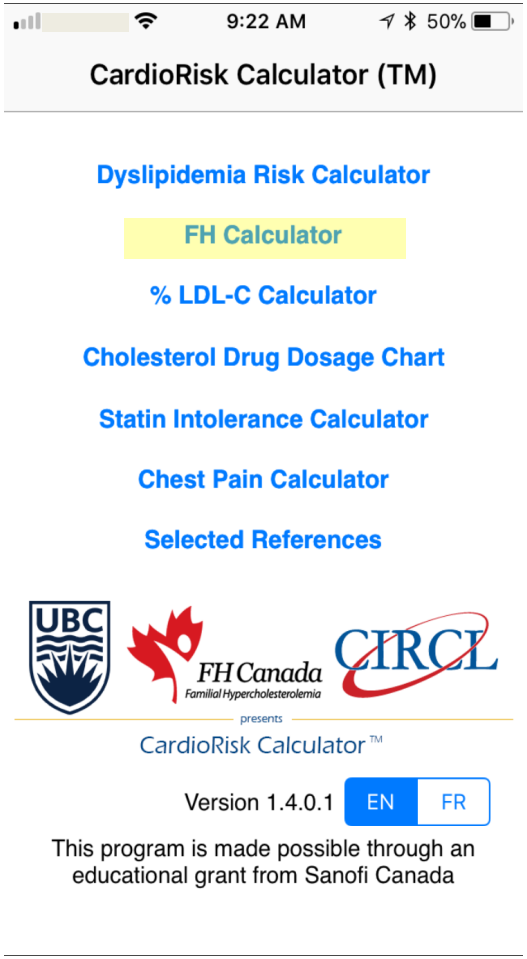
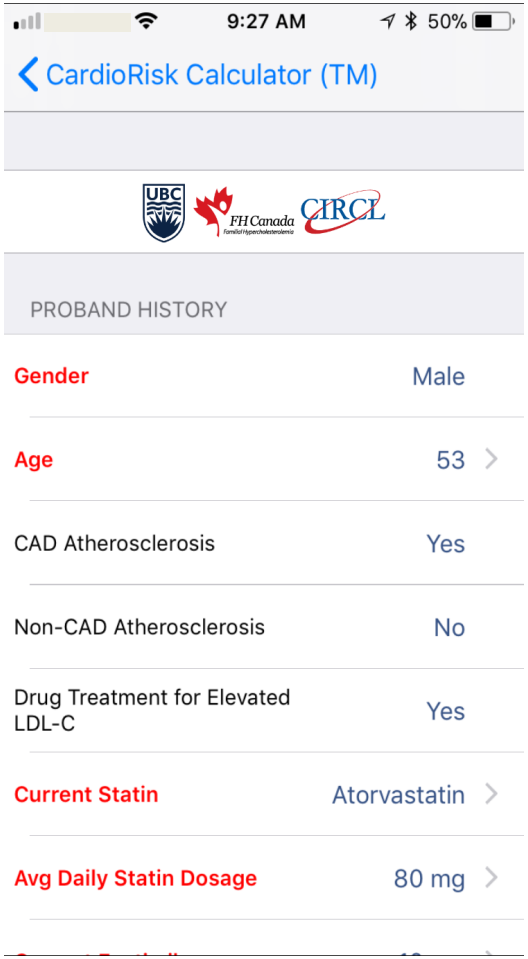
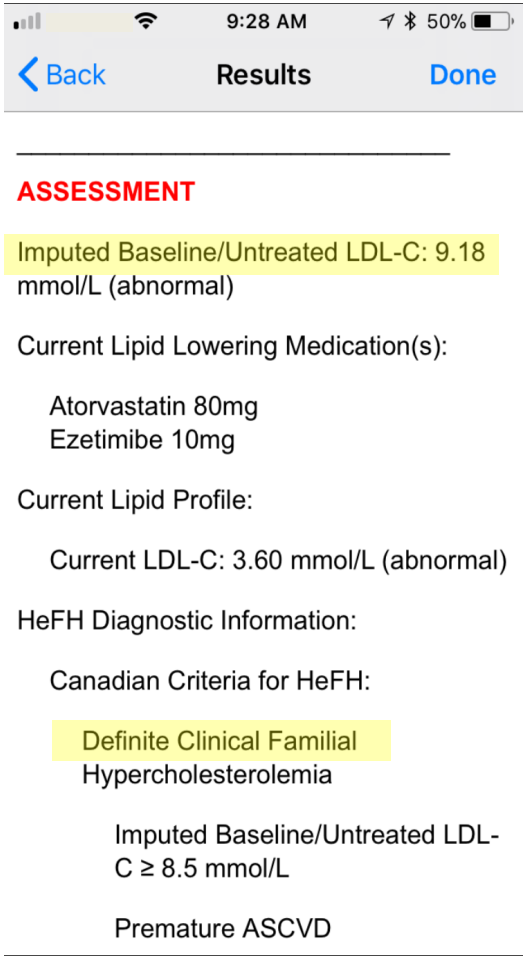






HF Canada
Hypercholestérolémie Familiale
FH Canada
Familial Hypercholesterolemia

6- Mise à jour de l'énoncé de position de la SCC sur l'HF

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

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]

[a] Department of Medicine, University of British Columbia, Vancouver, British Columbia, Canada; [b] Centre for Heart Lung Innovation, University of British Columbia, Vancouver, British Columbia, Canada; [c] Research Institute of the McGill University Health Centre, Royal Victoria Hospital, Montréal, Quebec, Canada; [d] Department of Medicine, McGill University, Montréal, Quebec, Canada; [e] Nutrition, Metabolism and Atherosclerosis Clinic, Institut de recherches cliniques de Montréal, Montréal, Quebec, Canada; [f] Faculty of Medicine, University of Toronto, Institute for Clinical Evaluative Sciences, Schulich Heart Centre, Sunnybrook Health Sciences Centre, Toronto, Ontario, Canada; [g] Mazankowski Alberta Heart Institute, University of Alberta, Edmonton, Alberta, Canada; [h] Department of Medicine, McMaster University, Hamilton, and Canadian Collaborative Research Network, Brampton, Ontario, Canada; [i] Departments of Medicine and Laboratory Medicine, CHU de Québec-Université Laval, Québec City, Quebec, Canada; [j] Centre for Heart Lung Innovation, University of British Columbia, Vancouver, British Columbia, Canada; [k] Department of Pediatrics, The Labatt Family Heart Centre, The Hospital for Sick Children, University of Toronto, Toronto, Ontario, Canada; [l] Department of Pediatrics, McMaster University, Hamilton, Ontario, Canada; [m] Department of Pediatrics, McGill University, Clinique 180, Montréal, Quebec, Canada; [n] Department of Pediatrics, Université de Montréal, CHU Sainte-Justine, Montréal, Quebec, Canada; [o] Departments of Medicine and Biochemistry, Schulich School of Medicine and Robarts Research Institute, Western University, London, Ontario, Canada; [p] Department of Medicine, University of Toronto and Division of Endocrinology, St Michael's Hospital, Toronto Ontario, Canada; [q] Lipidology Unit, Community Genomic Medicine Centre and ECOGENE-21, Department of Medicine, Université de Montréal, Saguenay, Quebec, Canada; [r] Department of Pathology and Molecular Medicine, Department of Clinical Epidemiology and Biostatistics, Population Health Research Institute and Thrombosis and Atherosclerosis Research Institute, Hamilton Health Sciences, McMaster University, Hamilton, Ontario, Canada; [s] Division of Endocrinology and Metabolism, Department of Medicine, University of Alberta, Edmonton, Alberta, Canada; [t] Queen Elizabeth II Health Sciences Centre, Dalhousie University, Halifax, Nova Scotia, Canada; [u] Department of Medicine, Division of Endocrinology, Université de Montreal, Montréal, Quebec, Canada; [v] Department of Medicine, Section of Endocrinology and Metabolism, University of Manitoba, St Boniface Hospital, Winnipeg, Manitoba, Canada; [w] Schulich School of Medicine and Dentistry, Western University, London, Ontario, Canada; [x] Division of Cardiology, Department of Medicine, Dalhousie University, St John, New Brunswick, Canada

Received for publication September 16, 2018. Accepted September 16, 2018.

[†]Deceased.

Corresponding author: Dr Liam R. Brunham, University of British Columbia Centre for Heart Lung Innovation, Rm 166 - 1081 Burrard St, Vancouver, British Columbia V6Z 1Y6, Canada. Tel.: +1-604-806-2344 ×63929; fax: +1-604-806-8351.
E-mail: Liam.brunham@ubc.ca

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.

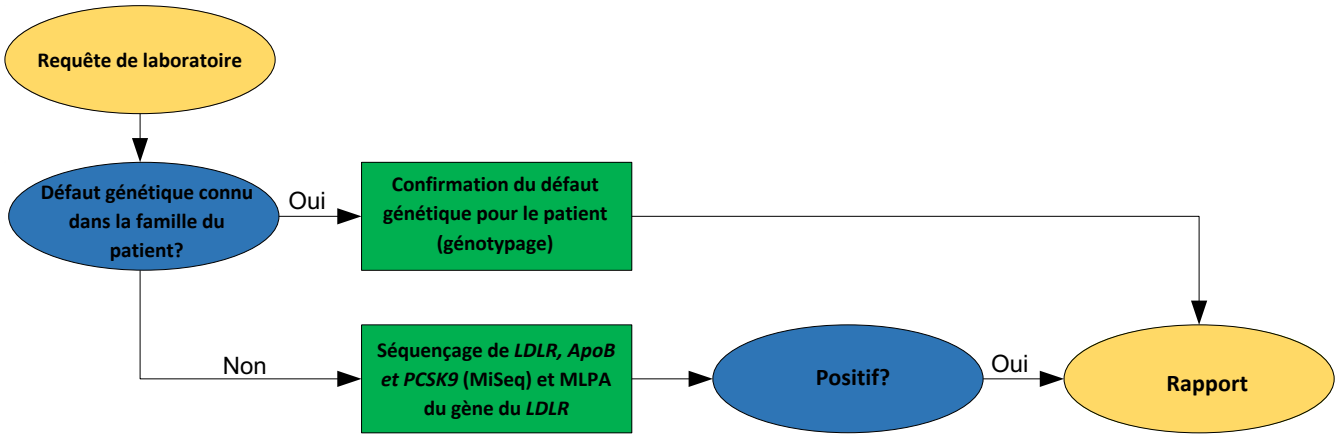
https://doi.org/10.1016/j.cjca.2018.09.005

HF Canada Hypercholestérolémie Familiale
FH Canada Familial Hypercholesterolemia

7- Mise en place d'un test génétique d'HF

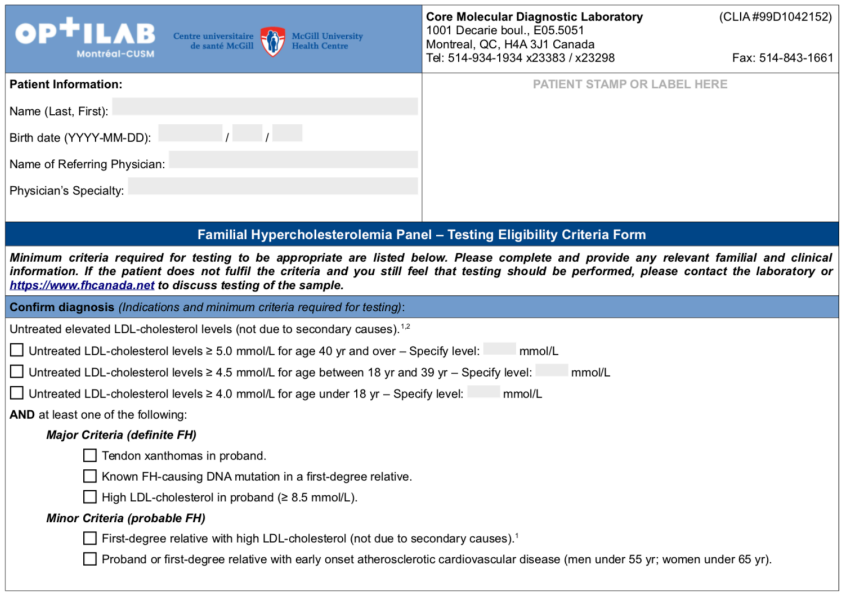
Le Laboratoire de diagnostique moléculaire - CMDL du Centre universitaire de santé McGill est le seul laboratoire de génétique clinique certifié CLIA pour le dépistage génétique de l'HF au Canada

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



HF Canada Hypercholestérolémie Familiale
FH Canada Familial Hypercholesterolemia

7- Mise en place d’un test génétique d’HF



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:

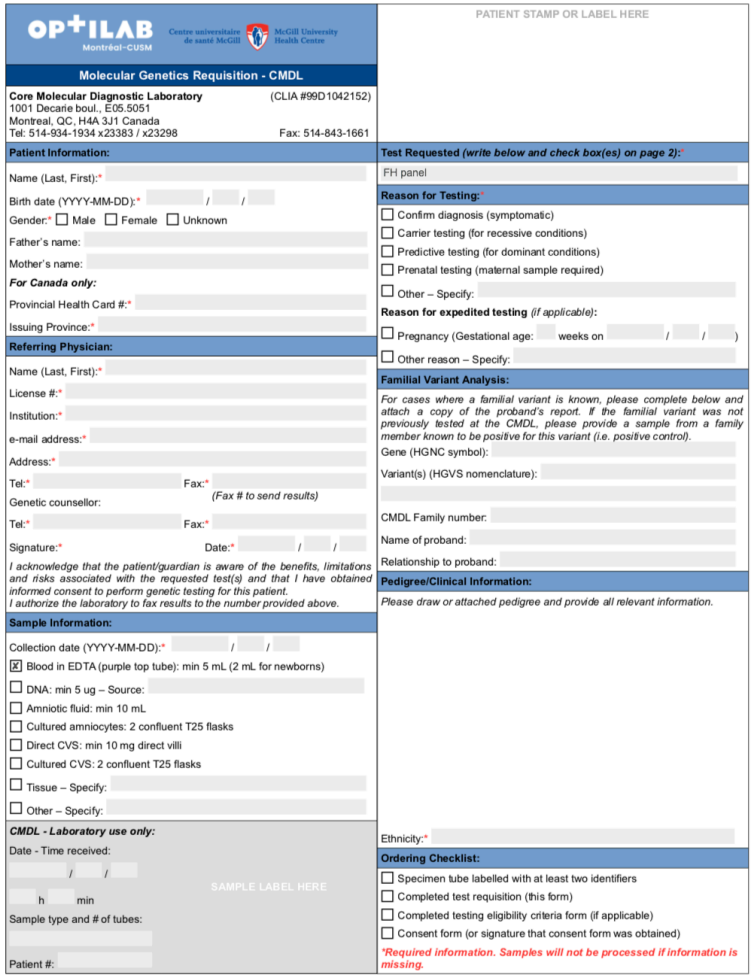
Major Criteria (definite FH)

- ☐ 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

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 STAMP OR LABEL HERE

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:

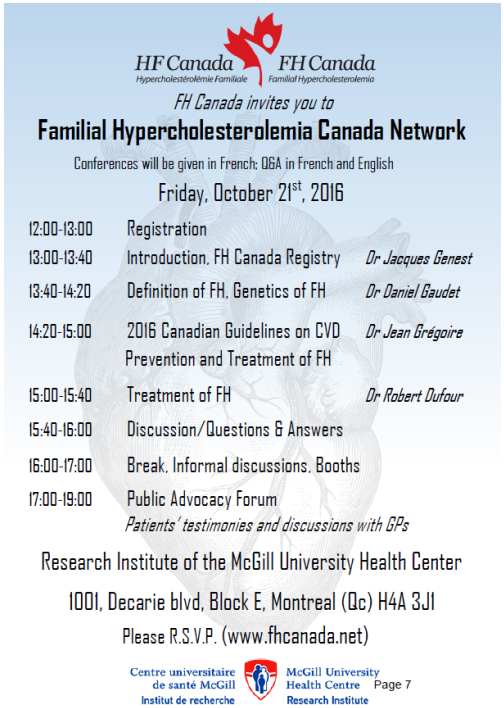
- ☐ Specimen tube labelled with at least two identifiers
- ☐ Completed test requisition (this form)
- ☐ Completed testing eligibility criteria form (if applicable)
- ☐ Consent form (or signature that consent form was obtained)

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

HF Canada Hypercholestérolémie Familiale — FH Canada Familial Hypercholesterolemia

Autres initiatives

Réseau HF Canada: réunions annuelles accréditées incluant un forum pour les patients.


HF Canada FH Canada
Hypercholestérolémie Familiale · 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

Page 7


CCRN Canadian Collaborative Research Network · HF Canada FH Canada

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
Vancouver, BC

FACULTY

Jacques Genest, MD, CM, FRCPC, FACC, FAHA
Professor and Novartis Chair in Medicine, McGill University
Scientific Director, Center for Innovative Medicine, McGill University Health Center/ Royal Victoria Hospital
Montreal, QC

Milan Gupta, MD, FRCPC, FACC
Associate Clinical Professor of Medicine, McMaster University
Assistant Professor of Medicine, University of Toronto
Medical Director, Canadian Collaborative Research Network
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
Scientist, UBC Centre for Heart Lung Innovation
Physician, Healthy Heart Program, Prevention Clinic, St. Paul's Hospital
Vancouver, BC

Milan Gupta
MD, FRCPC, FACC
Associate Clinical Professor of Medicine, McMaster University
Assistant Professor of Medicine, University of Toronto
Medical Director, Canadian Collaborative Research Network
Brampton, ON

PLANNING COMMITTEE

Jacques Genest
MD, CM, FRCPC, FACC, FAHA
Professor and Novartis Chair in Medicine, McGill University
Scientific Director, Center for Innovative Medicine, McGill University Health Center/ Royal Victoria Hospital
Montreal, QC

Peter J. Lin
MD, CCFP
Director Primary Care Initiatives
Canadian Heart Research Centre
Associate Editor
Elsevier WebPortal - Practice Update Primary Care
Medical Director LinCorp Medical Inc.
"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 FH Canada
Hypercholestérolémie Familiale · Familial Hypercholesterolemia

Conclusion: opportunités et défis

- Une meilleure compréhension des lacunes dans les soins pour les patients atteints d'HF au Canada
- Un ensemble simplifié de critères de diagnostic pour la population canadienne avec des outils qui facilitent le diagnostic de l'HF
- Un accès au diagnostic moléculaire d'HF
- Un accès à de nouveaux médicaments

Au fur et à mesure que la taille et la portée du registre national augmenteront, il sera possible d'améliorer le diagnostic et les soins de santé pour les patients avec HF au Canada.

HF Canada
Hypercholestérolémie Familiale
FH Canada
Familial Hypercholesterolemia

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FH Canada
Familial Hypercholesterolemia

Si vous avez des questions sur le registre, vous pouvez visiter notre site Web à l'adresse www.fhcanada.net

Vous pouvez également contacter la coordinatrice nationale:

Isabelle Ruel PhD
Institut de recherche du Centre universitaire de santé McGill
1001, boul. Décarie, E01.3144
Montréal, Québec
H4A 3J1
514-934-1934, poste 34852
info@fhcanada.net

McGill

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

UBC THE UNIVERSITY OF BRITISH COLUMBIA

Western UNIVERSITY · CANADA

UNIVERSITY OF TORONTO

Université de Montréal

Si vous avez des questions sur le registre, vous pouvez visiter notre site Web à l'adresse www.fhcanada.net

ou envoyer un courriel à
Isabelle Ruel, info@fhcanada.net

AMGEN

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