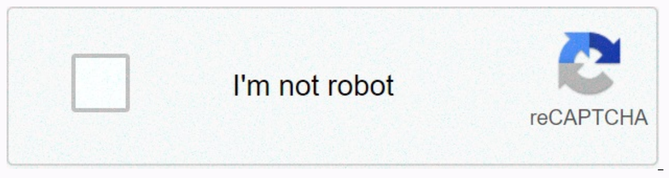

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Familial hypercholesterolemia canadian guidelines

Baixe o PDF aqui. Baixe a ferramenta ponto de cuidado para auxiliar a identificação de pacientes mais propensos a se beneficiar do encaminhamento para especialista em lipídios ou da revisão mais abrangente, o GEC-KO Messenger contendo mais sobre diagnóstico, triagem e gerenciamento e muito mais. Acesse um módulo de educação com aprendizado baseado em casos aqui. Resumindo: a hipercolesterolemia familiar é uma doença dominante autossômica comum (~1/250) que resulta em um aumento de 6 a 22 vezes em doenças cardiovasculares prematuras (DCV) e morte. O diagnóstico precoce e o tratamento podem normalizar a expectativa de vida. As principais características da hipercolesterolemia familiar são LDL-C elevado ≥ 5mmol/L com características adicionais, como cvd de início precoce (<55 years= in= men,=></55> <65 years= in= women),= cholesterol= deposition= in= the= tendons= (xanthomata)= and/or= around= the= eyes= (xanthelasma),= arcus= cornealis= with= onset=></65> <45years, and= family= history= of= early= onset= cvd= or= hyperlipidemia= requiring= treatment.= in= canada,= a= diagnosis= of= familial= hypercholesterolemia= is= typically= based= on= an= individual's= clinical= presentation= correlating= with= one= of= three= familial= hypercholesterolemia= definitions.= genetic= testing= is= not= generally= available,= and= a= clinical= diagnosis= guides= treatment= and= screening= of= family= members.= once= a= person= is= diagnosed= with= familial= hypercholesterolemia,= cascade= screening= of= family= members= using= measurement= of= ldl-c= levels= is= recommended.= this= enables= early= identification= and= treatment= of= at-risk= individuals ,= with= statins= as= first-line= treatment.= updated= sept= 2019= what= is= familial= hypercholesterolemia?= familial= hypercholesterolemia= (fh)= is= an= autosomal= dominant= genetic= condition= where= the= uptake= of= low-density= lipoprotein= cholesterol= (ldl-c)= into= cells= is= either= decreased= or= inhibited.= this= results= in= lifetime= exposure= to= very= high= levels= of= ldl-c.= fh= is= the= most= common= genetic= disorder= causing= premature= cardiovascular= disease= (cvd)= and= death= in= both= men= and= women.= about= 1= in= 250= canadians= is= thought= to= have= the= heterozygous= (hefh)= form= of= fh.= fh= is= more= common= in= certain= populations= due= to= founder= effects:= in= certain= areas= of= quebec,= the= prevalence= is= as= high= as= 1= in= 80;= it= affects= approximately= ~1/100= lebanese= and= afrikaners,= and= 1/67= south= african= ashkenazi= jews.= fh= is= under= diagnosed= and= under= treated= both= in= canada= and= worldwide ,= despite= the= knowledge= that= early= diagnosis= and= treatment= can= normalize= life= expectancy.= table= 1.= clinical= features= of= familial= hypercholesterolemia= in= heterozygotes= (hefh)= and= homozygotes= (hofh).= clinical= featureshefh= hofh= genetics= mutation= in= one= copy= of= one= fh= genemutation= in= both= copies= of= fh= gene= ldl-c= levels= ≥= 5mmol/l= with= additional= features= shown= in= following= boxes=>12 mmol/L lower LDL-C levels, especially in children or treated patients, do not exclude the onset of cardiovascular diseases hofh Other risks of atheosclerotic diseases: Stroke or transient isquemic attack — Peripheral vascular disease Physical findings — Cholesterol deposits in tendons (xanthomata) and/or around </45years,> of </45years,> (xanthelasma) — Arcus cornea (white, gray, or blue opaque ring on the cornea margin) family history - early-onset CVD — Hyperlipidemia, often requiring treatment AS FAMILY HYPERCHOLESTEROLEMIA IS DIAGNOSED? The Canadian Cardiovascular Society (CCS) recommends the use of canadian diagnostic criteria for FH proposed by the Family Hypercholesterolemia Canada (FHCanada) network (Figure 1). Although these criteria are relatively new, they are less complicated than those published by the Dutch Lipid Clinic Network (DLCNC) or the Simon Broome Registry (see the GEC-KO Messenger for more on these criteria) and have been validated against each of these criteria, which are internationally accepted for the diagnosis of HeFH. Genetic testing is not necessary for diagnosis, and are not routinely available in most of Canada. In Quebec, health professionals may request tests from the CHU Sainte Justine Molecular Laboratory focused on the most common genetic mutations found in French Canadians with family hypercholesterolemia Figure 1: Canadian criteria for the clinical diagnosis of family hypercholesterolemia (ESF). By Ruel I et al., 2018 Can J Cardiol. Reprinted with permission under the CC BY-NC-ND ASCVD license: athertotic cardiovascular disease; LDL-C: low-density lipoprotein cholesterol. * Secondary causes of high LDL-C should be ruled out (severe or untreated hypothyroidism, nephrotic syndrome, liver disease [biliary cirrhosis], medication, especially antiretroviral agents). CASCADING SCREENING The most economical approach to identifying new cases of family hypercholesterolemia is cascading screening of family members of the first individual with a confirmed diagnosis, known as an index case. This approach is recommended by the Canadian Cardiovascular Society (CCS). Screening may include lipid profiles of relatives and/or genetic testing for a known family mutation when available. Each newly diagnosed individual becomes a new case of index and cascading screening of relatives continues. Surveillance and MANAGEMENT Statins are the class of drugs chosen for individuals with HeFH. LDL-C should be reduced as quickly and as far as possible. The CcS recommends a reduction of >50% of LDL-C from the age of 18 years as primary prevention with the objective of LDL-C <2,0mmol/L for secondary prevention. Some individuals with FH will need combination and/or emerging therapy to obtain optimal LDL-C. Families with ESF should be advised about the importance of lifestyle modification, such as smoking cessation and avoiding passive smoking, diet, exercise, daily activity that starts early in life, maintaining ideal body weight and reducing stress. Children: The lifestyle changes discussed above continue to be the cornerstone of CVD prevention in children and adolescents with ESF and it is recommended to to a specialist for treatment decisions. See www.geneticseducation.ca for the comprehensive GEC-KO Messenger with full reference list and more Hofh, hofh. For the updated Canadian definition of FH visit www.FHCanada.net to find Ruel et al., simplified Canadian definition for family hypercholesterolemia. May J Cardiol 2018 34: 1210-1214. Authors: S Morrison MS CGC, JE Allanson MD FRCPC, RA Hegele MD FRCPC, S Hadjiyannakis MD FRCPC and JC Carroll MD CCFP GEC-KO in the race is for educational purposes only and should not be used as a substitute for clinical judgment. GEC-KO aims to assist the practicing physician by providing informed opinions about genetic services that have been rigorously developed and evidence-based. Physicians should use their own clinical judgment, in addition to the articles published and the information presented here. GEC-KO assumes no responsibility or liability resulting from the use of the information contained herein. Volume 30, Issue 12, December 2014, Pages 1471-1481See Summary By: John Sawdon Director of Education & Special Projects of the Cardiac Health Foundation of Canada Family Hypercholesterolemia is a genetic disorder characterized by very high plasma levels of low-density lipoprotein cholesterol (LDL-C) that increases cardiovascular risk by up to 20 times and causes early onset of cardiovascular disease. FH is the most common single genetic disease that leads to cardiovascular diseases. Often the ESF is not recognized until the individual has a cardiovascular event. If identified early and treated, life expectancy may be prolonged. If untreated, men will develop cardiovascular diseases in their thirties or forties and women approximately ten years later. The number of cases of Family Hypercholesterolemia is approximately 1 in 500, with French Canadians having a higher incidence rate at 1 in 275. Based on these figures, it is projected that 83,500 individuals in Canada have Family Hypercholesterolemia. (1) Most patients with family hypercholesterolemia are not diagnosed due to inconsistent screening practices and lack of awareness of diagnosis. The Canadian Cardiovascular Society recommended that the diagnosis of FH should rely on the Simon Broom Registry or the criteria of the Dutch Lipid Clinic network. (2) A) Simon Broom Registration Criteria Total cholesterol >7.5mmol/L (for adults) or >6.7mmol/L (for children under 16 years) low-density lipo LDL-C Protein cholesterol >4.9mmol/L (adults) or >4.0 mmol/L (for children under 16 years) A nodule or plaque in the patient tendon or any of the patients' first or second degree DNA relatives based on evidence of LDLR mutation or another ESF-related gene in the patient Family history of myocardial infarction before age 50 in a first or second degree relative or 60 years of age to relative first-degree Family history of total cholesterol >7.5mmol/L in any first or second grade B relative) Network of Dutch Lipid First-degree relative with premature cardiovascular disease or LDL-C >95 percentile, or personal history of premature peripheral or cerebrovascular disease or LDL-C between 155 mg/dL and 189mg/dL First grade with deposits of lips nodules in the tendon, corneal arcus, or Relative child of first degree less than 18 years with LDL-C in the historical 95 percentile or personal coronary artery disease LDL-C between 4.91 (190mg/dL) and 6.44 (249mg/dL) Presence of corneal arcus in a patient under 45 years of age LDL-C between 6.46 (2 50 mg/d L) and 8.51 mmol/L (329mg/dL) The presence of the LDL-C tendinous lipid nodule >8.53 mmol/L (330mg/dL) or MUTATION of the LDLR gene Figure 1 above reflects a single inherited gene compared to a double inherited gene in Family Hypercholesterolity. Figure 2 below reflects the passing on of this gene when considering first-degree relatives and second-degree relatives who are mentioned in the diagnosis of FH using both the criteria of the Simon Broom Registry and the criteria of the Dutch Lipid Clinic Network. Figure 2 the Family Hypercholesterolemia Family Tree Because Family Hypercholesterolemia is so difficult to diagnose and treat until someone has experienced a cardiac event, individuals who have relatives with high LDL-C readings or who have had a myocardial infarction at a young age are encouraged to register in FH Canada and engage in pharmaceutical and lifestyle therapy. The aim of the Registry is to improve the detection and management of individuals and families with ESF and other serious lipid diseases in Canada and lower high blood cholesterol and prevent early cardiovascular disease. The benefits of participating in the registry include: Early Diagnosis of Familial Hypercholesterolemia for the whole family Better management of high cholesterol levels Heart Disease Prevention in family members with FH Access to new medications as part of clinical trials(3) To learn more about registration and how it can benefit you and your family go to www.fhcanada.net or call Isabelle Ruel PhD at 1-514-934-1934 extension 34852. Figure 3 Chart analyzes the levels of Bad Cholesterol or LDL-C and Good Cholesterol HDL-C and Triglycerides When comparing blood cholesterol levels with the levels of those diagnosed with Family Hypercholesterolemia we find extremely high levels of Bad Cholesterol present that leads to atherosclerosis and subsequent myocardial infarction or heart attacks at a young age FH is a hereditary disease that leads to aggressive and premature cardiovascular diseases. This includes heart attacks, strokes and narrowing of heart valves. If the ESF is not treated, the estimated risk for a coronic event (4) is 50% for men up to 50 years of age and 30% for women at 60 years of age. This article aims to create awareness of Family Hypercholesterolemia. While we will discuss new treatment options in combating early cardiovascular disease during our next segment, current treatment options include substantial lyreducing LDL-C from Bad Cholesterol through the class of medicines known as statins pcsk9 inhibitors. (5) These drug treatments also include lifestyle management, such as how to in relation to diet, exercise, weight control, blood pressure control, diabetes control and smoking cessation. This includes special instructions for children from families identified with Family Hypercholesterolemia never to start smoking, given the huge risk of cardiovascular disease. For more information about Family Hypercholesterolemia, check out the references, register on www.fhcanada.ca or join the FH Canada Patient Network and sign up for your e-newsletter. Our next article will expand on treatment options, including the new class of drugs available and their clinical trial results. References: The articles, on the Website of the Cardiac Health Foundation of Canada, are presented with the understanding that the Foundation is providing information only and not providing medical advice. Consult your family doctor, specialist, or health care professional before implementing any of the ideas expressed in these articles. Articles.