Worldwide Hemochromatosis (Haemochromatosis)

References, Tips and Websites

© 2019 George R Lewycky http://georgenet.net/bloodhemo grlewycky [at] yahoo dot com

#iron #phlebotomies #exjade #organizations #clinicaltrials #fda #glossary #livertoxicity #genetics #finding-a-doctor #tips #tests #Hemochromatosis #Haemochromatosis #liver #symptoms

FEEL FREE TO EMAIL ME OTHER WEBSITES OR INFO YOU THINK WOULD BENEFIT OTHERS HERE

<script type="text/javascript"

src="//rf.revolvermaps.com/0/0/6.js?i=5e3ilf7jle4&m=7&c=e63100&cr1=ffffff&f=arial&l=0&a
mp;bv=90&lx=-420&ly=420&hi=20&he=7&hc=a8ddff&rs=80" async="async"></script>

History & Genetics

The disease was first described in 1865 by Armand Trousseau in a report on diabetes in patients presenting with a bronze pigmentation of their skin which is why it's also called "*Bronzed diabetes*".

The term "haemochromotosis" was first used in 1889 by Friedrich Daniel von Recklinghausen <u>https://en.wikipedia.org/wiki/Friedrich_Daniel_von_Recklinghausen</u> describing the dark tissue staining in the liver now known to be from heavy deposits of haemosiderin leading to cirrhosis and organ damage.

A human has 23 pairs of chromosomes--a set from each parent. In the 1970s, French researchers first mapped the gene for hemochromatosis to the vicinity of the human lymphocyte antigen (HLA) region on the short arm of chromosome 6, in which 85% of the 178 patients diagnosed with hemochromatosis. The iron overloading disease has the same mutation in both pairs

Dr. John Feder and a team of scientists including Dr. Roger Wolff of Mercator Labs Menlo Park, California (also known as Progenitor Labs) discovered the gene for hemochromatosis, named it **HFE** (*h* for hemochromatosis, and *fe* for iron) in August 1996.

http://www.the-scientist.com/?articles.view/articleNo/18979/title/Genetics/ http://articles.latimes.com/1996-07-30/news/mn-29419_1_iron-overloading http://www.nature.com/ng/wilma/v13n4.868281725.html http://www.pubfacts.com/author/Roger+K+Wolff

http://www.bioiron.org/pdf/nomenclature%202.pdf

- Juvenile hemochromatosis. This affects adolescents and young adults ages 15 to 30. This form leads to a severe overload of iron, which can cause heart and liver disease.
- **Neonatal hemochromatosis.** In this form, iron builds up very quickly in an infant's liver and can cause severe organ damage.
- **Transfusion related hemochromatosis**. This is a result of iron overload caused by many blood transfusions in patients with disorders that cause them to not make enough red blood cells.

Myths & Realities

Famous people with HH

http://blogcritics.org/even-celebrities-are-not-immune-to/

- The Hemingway's: Ernest, his siblings Ursula and Leicester, his father Clarence, and his granddaughter Margaux
- John Steinbeck
- Steve (Terrence Steven) McQueen
- John Vernon Bouvier III (father of former First Lady Jacqueline Kennedy Onassis)
- Marie Warder
- Beethoven (cirrhosis due to hemochromatosis?)
- Jim Becker (Green Bay Packers)
- Frederick Abinger (Tom) Warder

Youtube videos

Hemochromatosis videos on Youtube

https://www.youtube.com/playlist?list=PLyw5PCdDWDO5q_piCXWkoo2MNGF14SsES

Anemia & Hematology videos on Youtube

https://www.youtube.com/playlist?list=PLyw5PCdDWDO683AuABWc7YZusWUmFSazh

Related books on Amazon

https://www.amazon.com/hz/wishlist/ls/EEP2411VXD4C?ref_=wl_share

<u>Twitter</u>

Twitter resources and news

https://twitter.com/search?src=typd&q=hemochromatosis

- https://twitter.com/Haem
- https://twitter.com/haemchrtosisIRL https://twitter.com/HepCommJournal
- https://twitter.com/IrishHaemAssoc
- https://twitter.com/MolecPath
- https://twitter.com/PKdeficiency

International mailing list of patients provided by Iron Disorders Institute

http://www.irondisorders.org/online-discussion-group/

Phlebotomies & Locations

http://www.hemoinfo.org/facilities/

<u>http://www.fda.gov/BiologicsBloodVaccines/BloodBloodProducts/RegulationoftheBloodSupply/Variances/ucm164649.h</u> <u>tm</u>

https://www.fda.gov/vaccines-blood-biologics/regulation-blood-supply/list-establishments-granted-approval-variance-21cfr6403d-and-21cfr6403f

http://www.toomuchiron.ca/2014/04/therapeutic-phlebotomy-techniques-a-patients-view/ https://en.wikipedia.org/wiki/List_of_blood_donation_agencies_in_the_United_States https://en.wikipedia.org/wiki/Blood_donation

FDA Collection of Blood and Blood Products from Patients with Hemochromatosis

https://www.fda.gov/BiologicsBloodVaccines/BloodBloodProducts/RegulationoftheBloodSupply/Variances/ucm164649. htm

Donating Blood

Can I donate blood or will a blood bank accept my blood instead of me going for a phlebotomy ? <u>http://abcnews.go.com/Health/MensHealth/football-fan-paying-tickets-saved-fatal-disease/story?id=10109129</u> <u>http://www.ncbi.nlm.nih.gov/pubmed/15190395</u> <u>http://www.redcrossblood.org/donating-blood/eligibility-requirements/eligibility-criteria-topic</u>

Blood tests & calculations

https://www.omnicalculator.com/health/transferrin https://www.omnicalculator.com/health/bmi

CHELATION, Exjade & Phlebotomies alternatives

http://georgenet.net/bloodhemo/FILES/images/Phlebotomy%20alternatives.jpg http://www.aferrix.com/PDF/Porter_DFO_DFX_2013.pdf http://www.irondisorders.org/chelation-therapy http://www.thalassemia.com/treatment-chelation.aspx#gsc.tab=0 http://www.thalassemiapatientsandfriends.com/index.php/board,5.0.html http://www.us.exjade.com/index.jsp

Clinical Trials

techniques-a-patients-view/

http://hemonc.cho.org/media/2015-sickle-cell-research-studies.pdf https://clinicaltrials.gov/ https://clinicaltrials.gov/ct2/about-studies/learn https://clinicaltrials.gov/ct2/results?term=hemochromatosis&Search=Search http://navigator.reaganudall.org/

Iron

http://www.anemiainwomen.com/html/about_iron.htm http://www.melbournehaematology.com.au/pdfs/guidelines/melbourne-haematology-guidelines-iron-studies.pdf http://www.merckmanuals.com/home/blood-disorders/iron-overload/hemochromatosis http://www.webmd.com/a-to-z-guides/iron-fe https://en.wikipedia.org/wiki/Iron_overload https://getpocket.com/explore/item/iron-is-the-new-cholesterol blood transfusions (http://www.nhlbi.nih.gov/health/health-topics/topics/bt **Certain medications can absorb Iron also** http://www.toomuchiron.ca/2014/04/therapeutic-phlebotomy-

Transplants or being a donor

https://unos.org/ https://organize.org/

https://www.research.va.gov/research_in_action/First-successful-liver-transplant.cfm

https://transplant.surgery.ucsf.edu/conditions--procedures/liver-transplant.aspx

https://www.mayoclinic.org/tests-procedures/liver-transplant/about/pac-20384842

https://liverfoundation.org/for-patients/about-the-liver/the-progression-of-liver-disease/liver-transplant/

https://columbiasurgery.org/liver/faqs-about-life-after-liver-transplant

https://my.clevelandclinic.org/health/treatments/8111-liver-transplantation

https://www.nyp.org/transplant/transplant-services/liver-transplant

http://www.childrenshospital.org/conditions-and-treatments/treatments/liver-transplant

https://www.uchicagomedicine.org/conditions-services/transplant/liver-transplant

https://www.yalemedicine.org/conditions/liver-transplant/

Rare Disorders

https://rarediseases.org National Organization for Rare Disorders https://rarediseases.org/contact-us/ https://www.raredisorders.ca/ (Canada CORD) https://rarediseases.org/ (United Stated NORD) https://www.raredisorders.ca/resources/ Assorted International organizations

Liver Glossary

http://www.globalliver.org/liver-glossary https://livertox.nih.gov/glossary.html

Children's Liver Association

http://samples.sainsburysebooks.co.uk/9781444300543_sample_383161.pdf

http://www.childrenshospital.org/conditions-and-treatments/treatments/liver-transplant

<u>https://www.classkids.org/</u> American Liver Foundation: <u>www.liverfoundation.org</u> CORE- Center for Organ Recovery & Education: <u>www.core.org</u> Children's Hospital of Pittsburgh: <u>http://www.chp.edu/our-services/transplant/liver/liver-transplant-necessary/liverdiseases-transplant</u>

Are your medications (past & present) safe on your liver

Liver Toxicity database of Medications (DILI/DILD) - see if your medications past or present could be causing liver damage

https://livertox.nih.gov/ https://livertox.nih.gov/drugliverinjury.html

Drug Induced Liver Injury (Disease) DILI / DILD

http://www.hepcnet.net/drugsandliverdamage.html http://www.liverfoundation.org/downloads/alf_download_614.pdf https://www.merckmanuals.com/home/liver-and-gallbladder-disorders/drugs-and-the-liver/effects-of-drugs-on-theliver

Medications, Side effects and Dictionary

 https://www.drugs.com/
 can create your own account to track past and present meds

 https://www.rxlist.com

 https://www.rxlist.com/drug-medical-dictionary/article.htm

 https://www.fda.gov/drugs/drug-information-consumers/finding-and-learning-about-side-effects-adverse-reactions

ORGANIZATIONS - WORLDWIDE

Hemochromatosis Information Society http://www.hemoinfo.org/ http://www.hemoinfo.org/facilities/

Gastroenterological Society of Australia https://www.gesa.org.au/resources/patients/health-information-fact-sheets/ https://www.gesa.org.au/resources/reference-links/ https://twitter.com/HepCommJournal

British Liver Trust https://www.britishlivertrust.org.uk/liver-information/liver-conditions/haemochromatosis/

The Haemochromatosis Society (Haemochromatosis UK) <u>http://haemochromatosis.org.uk/</u> http://haemochromatosis.org.uk/resources/

Irish Haemochromatosis Association <u>http://www.haemochromatosis-ir.com/home.html</u> <u>http://www.haemochromatosis-ir.com/other-associations-and-societies</u>

Global Liver Institute <u>http://www.globalliver.org/resources</u> Liver Disease Resources <u>https://twitter.com/GlobalLiver</u> Hepatoma Research <u>https://hrjournal.net/</u> <u>https://twitter.com/HepatomaRes</u>

Canadian Hemochromatosis Society (CHS)

https://www.toomuchiron.ca/ https://www.blood.ca/en/blood/iron https://www.toomuchiron.ca/support/chapters/ https://twitter.com/IronOutCanada

International BioIron Society http://www.bioiron.org/default.aspx http://www.bioiron.org/resources/links.aspx

Haemochromatosis UK

http://haemochromatosis.org.uk/ https://www.facebook.com/groups/haemochromatosis/

<u>Irish</u>

Irish Haemochromatosis Association – Dublin: http://www.haemochromatosis-ir.com/

IRISH DNA ATLAS PROJECT

https://www.familyhistory.ie/index.php/en/2012-04-22-23-12-47 https://www.familyhistory.ie/docs/DNA/DNA_01.pdf http://www.americanhs.org/irish%20in%20the%20blood.htm Celtic Curse http://celticcurse.org/ https://twitter.com/CelticCurse

Haemochromatosis Australia

http://www.haemochromatosisza.org/ http://www.melbournehaematology.com.au/ https://haemochromatosis.org.au/ https://haemochromatosis.org.au/links/ https://haemochromatosis.org.au/wordpress/wp-content/uploads/2018/04/HYQA2017-web.pdf https://twitter.com/Haem

UNITED STATES ORGANIZATIONS

American Hemochromatosis Society http://www.americanhs.org/ http://www.americanhs.org/faq.htm https://twitter.com/testhfe

Iron Disorders Institute (U.S.) www.irondisorders.org

National Heart, Lung, and Blood Institute (U.S.) www.nhlbi.nih.gov American Liver Foundation (ALF) http://www.liverfoundation.org

American Association for the Study of Liver Diseases https://www.aasld.org http://americanhs.org https://twitter.com/AASLDtweets

Iron Disorders Institute

http://www.irondisorders.org/ https://www.hemochromatosis.org/ https://www.hemochromatosis.org/connect-with-us/

Genetics & finding a Genetics Counselor (USA & Canada)

Geneticist / Genetics Counselor - maybe if planning on children or need medical clarifications

(US) <u>http://www.nsgc.org/page/find-a-gc-search</u>

(Canada) https://www.cagc-accg.ca/index.php?page=225&id=

(Glossary) https://www.genome.gov/glossary/index.cfm

http://www.bioiron.org/pdf/nomenclature%202.pdf

http://consumerhealthchoices.org/wp-content/uploads/2015/10/ChoosingWiselyGeneticTestsACMG-ER.pdf

Navigating the Genetics of Iron Overload

http://rickssnp.weebly.com/doctors.html | http://rickssnp.weebly.com/genetics-terms.html

The Hemochromatosis Gene <u>http://ironic-health.com/</u>

LINKS TO FIND A DOCTOR

Links below can help you find doctors based on your city, zipcode, insurance, etc.

https://health.usnews.com/doctors https://www.castleconnolly.com/ https://www.consumerreports.org/doctors/how-to-find-a-good-doctor/ https://www.healthgrades.com/ https://www.zocdoc.com/ https://amino.com/find-care/

Finding a Hemochromatosis Doctor

http://www.americanhs.org/doctors.htm http://celticcurse.org/wp-content/uploads/hemo-doc-stars-list-7-2014.pdf

http://www.lifescript.com/doctor-directory/condition/h-hereditary-hemochromatosis.aspx

Expertscape Ranks Top Doctors and Hospitals in Hemochromatosis <u>http://www.expertscape.com/ex/hemochromatosis</u> <u>https://www.prlog.org/12273488-expertscape-ranks-top-doctors-and-hospitals-in-hemochromatosis.html</u>

Doctors

What doctor(s)/specialists should I see or might be involved with?

- Gastroenterologist
- Hepatologist liver specialist
- Endocrinology diabetes is common with HH

- Hematologist/Oncologist study and analyze your bloodwork; genetics; phlebotomies (removing blood to lower the Iron in your blood stream)
- Nutritionist/Dietician to remove Iron out of the your diet and increase other nutrients you might need
- Pathologist reviews MRI, CAT, Ultrasound or Biopsy
 - Might be beneficial to get 2nd opinions or ask the doctor for another review
- Geneticist / Genetics Counselor maybe if planning on children or need medical clarifications
 - (US) <u>http://www.nsgc.org/page/find-a-gc-search</u>
 - (Canada) <u>https://www.cagc-accg.ca/index.php?page=225&id=</u>
 - (Glossary) <u>https://www.genome.gov/glossary/index.cfm</u>
- Consider 2nd opinions or even firing your doctor and re-evaluating test results if you are not satisfied

Check to see if the doctor has experience with Hemochromatosis, phlebotomies, liver diseases.

As always keep copies of your medical tests and records, remember Doctors move, retire, pass away, etc.

Teaching hospitals are a good place to start and they would all be on the same computer/records system also

Common Tests/Diagnostics

- Blood Tests
 - <u>https://labtestsonline.org/_g</u>ood reference of values of many blood tests that are done
 - 0
 - Standard
 - Liver Function Tests (LFT) <u>http://www.aafp.org/afp/1999/0415/p2223.html</u>
 - Complete Blood Count (CBC)
 - Ferritin over 200-300 ng /ml is probably hemochromatosis
 - Ferritin reflects the body's iron stores. Blood ferritin levels increase when the body's iron stores increase.
 - https://medlineplus.gov/ency/article/003490.htm
 - •
 - Transferrin saturation (**TSAT**)
 - Transferrin is a protein that binds and transports iron around the body. With higher levels
 of iron, more of the transferrin is carrying iron. Transferrin saturation rates of 45 percent
 should be evaluated.
 - (transferrin saturation index) = serum iron / TIBC
 - Try to keep between 20- 50%
 - o **Hgb**
 - ALT <u>http://www.gastroendonews.com/Hepatology-in-Focus/Article/08-17/ACG-Redefines-</u> Normal-Range-of-ALT/42158
 - 0

0

- MCV if low probably iron deficiency until hematologist reviews case.
 - Iron deficiency says they are bleeding until some good gastroenterologist
 - Indicator of bone marrow is a rich dark-maroon color laden with iron or a dull gray victim of piracy from occult bleeding, malabsorption, genetic sabotage, or dietary neglect
- o Iron panel
- Serum Iron <u>https://medlineplus.gov/ency/article/003488.htm</u>
- 0
- o serum ferritin and/or gamma-glutamyl transpeptidase (GGT) levels
- Additional
 - Vitamin D deficiency very common
 - <u>http://articles.mercola.com/sites/articles/archive/2014/05/28/vitamin-d-deficiency-signs-symptoms.aspx</u>

- •
- B12 red cells reproduction especially if anemic or Hgb drops
- NOTE: some laboratories have different ranges and they can be computed on age, gender
- Ultrasound (US)
 - o Don't take Pathologist report/findings as gospel, might need MRI
- MRI (w/wo contrast)
- MRI w/ Ferriscan accurate measurement of liver iron concentration (LIC) through a non-invasive, MRIbased technology
 - <u>http://www.resonancehealth.com/products/ferriscan-mri-measurement-of-liver-iron-concentration.html</u>
 - <u>http://www.resonancehealth.com/images/files/FerriScan/FerriScan%20Fact%20Sheet%20Mar%20</u> 2015.pdf
 - http://haemochromatosis.org.au/wordpress/wp-content/uploads/2012/12/Ferriscan-Fact-Sheet-Web.pdf
 - o <u>https://twitter.com/hashtag/ferriscan</u>
- **Bone marrow aspiration** is a procedure that involves taking a sample from the soft tissue inside your bones. Bone marrow is the spongy tissue found inside bones. It contains cells that produce white blood cells, red blood cells, and platelets inside larger bones such as the: spine and *invasive*
- •
- Bone marrow aspiration (biopsy): the procedure is similar to liver biopsy in that a needle is used to penetrate the bone marrow. The needle gathers a small amount of tissue from the marrow, which is stained for iron in the same way as a liver biopsy. Bone marrow tissue reveals the blood cell health and activity. This procedure is performed most often on patients with blood cancers, unexplained iron deficiency anemia or who are preparing for bone marrow transplantation. (*invasive & gold standard but risky*)
 - http://www.mayoclinic.org/tests-procedures/bone-marrow-biopsy/basics/why-its-done/prc-20020282
 - o <u>http://www.pathologyoutlines.com/topic/bonemarrowiron.html</u>
- Fibroscan to see if Iron is in your liver besides your bloodstream. non-invasive technology
 - o http://www.myliverexam.com/en/lexamen-fibroscan.html
 - <u>http://www.fibroscan.com/en/products</u>
 - 0
- CT (computed tomography) -
- Genetics (HFE: C282Y or H63D are the most common. S65C & 845A also exist)

 LAL-D
- Bone Density test especially if Vitamin D deficient, Osteoporosis
- Ferritometer or SQUID for Superconducting Quantum Interference Device—is a non-invasive technology
 that measures liver iron in about 15 minutes using super-cooled magnets. Uses magnetic fields to measure
 the amount of iron stored in the liver. Non-invasive liver iron measurements are currently available at two
 locations in the United States: Columbia-New York Presbyterian in New York (SQUID) and Children's
 Hospital & Research Center in Oakland, California.
 - http://www.indianpediatrics.net/apr2012/apr-268-269.htm
 - o <u>http://www.tristantech.com/</u>
 - o <u>http://www.irontoxicity.com/hcp/diagnosis/imaging_studies/magnetic.jsp</u>
 - 0
 - UCSF Benioff Children's Hospital Oakland has one of only four ferritometers in the world. <u>http://www.thalassemia.com/research-squid.aspx#gsc.tab=0</u>
 - o <u>http://www.childrenshospitaloakland.org/main/news/204.aspx</u>
 - o http://www.chori.org/Current_News/Previous_Years/01_feb_ferritometer.html
- Liver Biopsy the gold standard and *invasive*
 - <u>http://www.hopkinsmedicine.org/gastroenterology_hepatology/clinical_services/specialty_service</u> <u>s/liver_biopsy.html</u>
 - Liver stores 70% of body's iron
 - Liver biopsy will help confirm the diagnosis and identify the amount of scarring, if any, in the liver.
 - Don't take Pathologist report/findings as gospel
 - Can request your slides be reviewed by another pathologist (2nd opinion)

- **Aixplorer** the first ultrasound system to provide you with the following imaging benefits of ShearWave Elastography which is a comfortable, non-invasive technology that can be used for assessing and staging patients with chronic liver disease
 - <u>http://www.supersonicimagine.com/Aixplorer-R/Hepatology-Gastroenterology</u>
 - o <u>http://www.sandhillsci.com/wp-content/uploads/2016/03/Sandhill-Aixplorer-Brochure.pdf</u>

FIRST DOCTOR VISIT

What do I tell my doctor ?

Ever had any blood transfusions - could explain iron

Family Heritage / Genetics – your last name might not be obvious for them to detect if Irish, Italian, etc that usually have HH

Medications & Supplements you take past and present

Especially medication that are known to damage your liver

https://www.merckmanuals.com/home/liver-and-gallbladder-disorders/drugs-and-the-liver/effects-of-drugs-on-the-liver

Tattoos even

USUAL DOCTOR EXAM / RE-EXAM

Blood test

Checking Thyroid and ankles

Examine liver area (fatty liver, etc)

Decide if you need a phlebotomy (maintenance)

Review you medications (past & present) especially highlight medications listed here:

http://www.hepcnet.net/drugsandliverdamage.html

https://www.merckmanuals.com/home/liver-and-gallbladder-disorders/drugs-and-the-liver/effects-of-drugs-on-the-liver

http://drugs.com has a good feature that's free to maintain your medications and print it when needed

Family & Medical history and/or heritage and genetics (Last name/Married name misleading)

Drinking alcohol

Ask if vaccines needed: Hepatitis A & B?

Any Liver issues currently, or family history (cancer, hepatitis, etc)

http://www.mayoclinic.org/diseases-conditions/hemochromatosis/diagnosis-treatment/preparing-forappointment/ptc-20167333

Images of HH, Organs affected, Liver

http://georgenet.net/bloodhemo/FILES/ FirstTimers/OrgansAffectedIron-Overload.jpg http://georgenet.net/bloodhemo/FILES/ FirstTimers/Hepatic_PhysicalSigns.jpg http://georgenet.net/bloodhemo/FILES/ FirstTimers/Diagram_ChronicLiverDisease.jpg http://georgenet.net/bloodhemo/FILES/images/hemochromatosis.jpg http://georgenet.net/bloodhemo/FILES/images/Skin%20bronzing.jpg http://georgenet.net/bloodhemo/FILES/images/liverdiagram2.jpg

Images of NASH & NAFLD

http://georgenet.net/bloodhemo/FILES/images/liverstages_NASH.jpg http://georgenet.net/bloodhemo/FILES/images/NASH%20%26%20NAFLD.png http://georgenet.net/bloodhemo/FILES/images/NAFLD_Nature.jpg

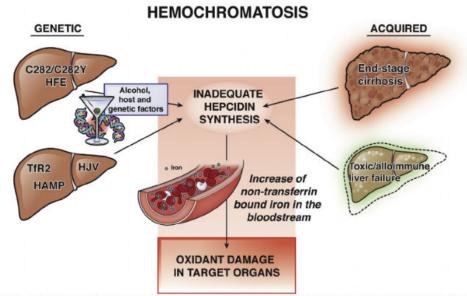
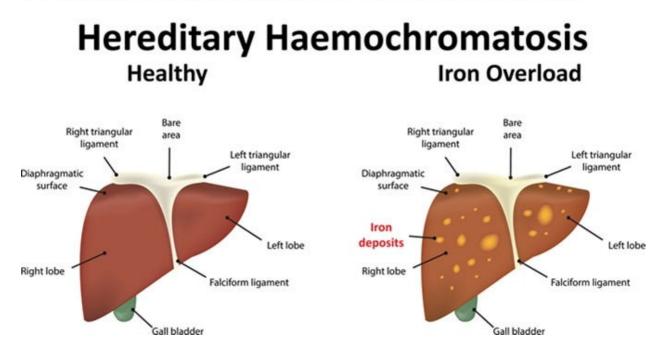
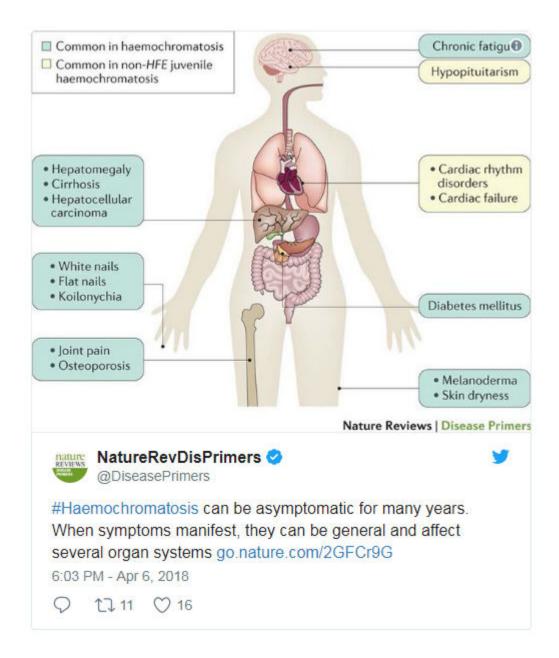


Figure 5. Genetic and nongenetic causes of hemochromatosis. The clinical syndrome of hemochromatosis, characterized by inappropriate hepcidin synthesis/activity, progressive appearance in the serum of non-transferrin-bound iron, including redox-active forms, such as labile plasma iron, followed by tissue iron overload in parenchymatous organs is associated with genetic loss of proteins involved in hepcidin regulation (genetic or hereditary hemochromatosis) or hepatic disorders leading to persistent and prolonged loss of hepcidin-producing liver mass (acquired hemochromatosis) (see text for details).





By George R Lewycky

Grlewycky [at] yahoo dot com

http://Georgenet.net/bloodhemo

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