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Sullivan Nicolaides PATHOLOGY
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New lipid reporting parameters expand asCVD risk guidance

By **Dr Gemma Daley** MBBS(Hons 1st Class) **MBA MAACB**

As part of our lipid profile, we will now be reporting non-HDL cholesterol, an important marker for atherosclerotic cardiovascular disease (asCVD) risk, perhaps even superior to LDL-cholesterol.

Key points

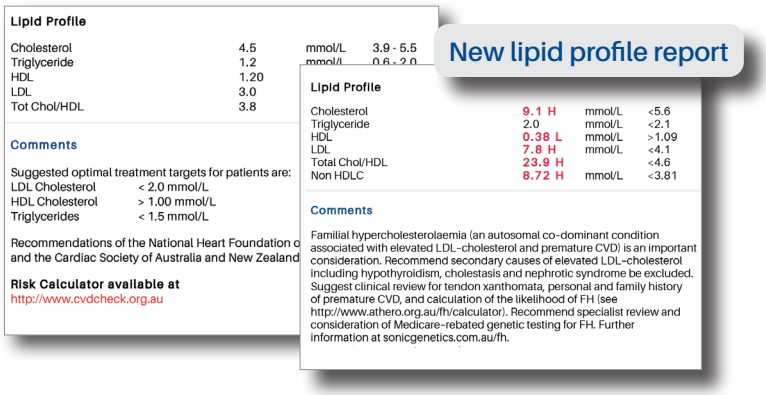
- for most patients a non-fasting specimen is preferred for assessment of lipids
- Familial Hypercholesterolaemia is an important and underdiagnosed condition
- Sonic Genetics is now offering genetic testing for Familial Hypercholesterolaemia
- a Medicare rebate for genetic testing for Familial Hypercholesterolaemia was introduced in May 2020

Non-fasting specimens are preferred

Traditionally, pathology providers have requested patients fast before having their lipids measured. For many of our patients, fasting is inconvenient and can act as a barrier to having their blood taken. Furthermore, patients with diabetes are at risk of hypoglycaemia should they fast.

We know that lipids change minimally in the non-fasting state, removing the requirement for fasting samples. Since most people are in the non-fasting state for the majority of the day, a non-fasting lipid profile is a more accurate reflection of their asCVD risk.[1]

There is a small group of patients where a fasting sample may be required.



Non-HDL cholesterol preferred for risk prediction

Non-HDL cholesterol can give valuable information about a patient's risk for asCVD. It is a calculation based on total cholesterol and HDL-cholesterol. Experts have suggested that it may well be superior to LDL-cholesterol for asCVD risk prediction, particularly in patients with high triglycerides, and in those with very low LDL-cholesterol concentrations.[2] We will now provide non-HDL cholesterol with our usual lipid report. Non-fasting specimens are again preferred.

What this means for your patients

These changes and advancements in lipid reporting have the following benefits:

- Patients may have their blood test at a time that suits them during the day
- Patients with diabetes are not at risk of hypoglycaemia from fasting
- Non-HDL cholesterol reporting will improve our assessment of a patient's asCVD risk
- if eligible, patients will have access to a Medicare rebate for genetic testing for familial hypercholesterolaemia.

Familial Hypercholesterolaemia

Familial Hypercholesterolaemia (FH) is an underdiagnosed genetic condition thought to affect 1 in 250 Australians. FH is important because it is associated with coronary and peripheral vascular disease, particularly if left untreated.[3] Comments will now be provided with the lipid profile to indicate when the risk of FH is significantly increased. An individual's risk can then be calculated using the Dutch Lipid Clinic Network Score. See over for more information on FH genetic testing.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Further Information

Dutch Lipid Clinic Network Score
https://www.athero.org.au/fh/wp-content/uploads/Dutch-Lipid-Clinic-Network-Score2.pdf

Familial Hypercholesterolaemia (Sonic Genetic Pathology Educational Video)
https://www.soniceducation.com.au/clinical-resources/online-videos/genetic-pathology/

Sonic Genetics Hypercholesterolaemia Panel
https://www.sonicgenetics.com.au/our-tests/all-tests/hypercholesterolaemia-panel/

References:
1. Langsted & Nordestgaard. Nonfasting Versus Fasting Lipid Profile For Cardiovascular Risk Prediction. Pathology 2019;51(2):131-41.
2. Carr S, et al. Non-HDL-cholesterol and Apolipoprotein B Compared With LDL-cholesterol in Atherosclerotic Cardiovascular Disease Risk Assessment. Pathology 2019;51(2):148-54.
3. Pang J, Sullivan DR, Brett T, Kostner, KM, Hare DL, Watts GF. Familial Hypercholesterolaemia in 2020: A leading tier 1 genomic application. Heart, Lung Circ 2019.doi.org/10.1016/j.hlc.2019.12.002

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Patient test collection notes

Updated and new instructions

SNP provides collection notes to assist patients to prepare for pathology collections. These notes can be supplied to your clinic as a full set and re-ordered using the pathology stores request form. Collection notes can also be downloaded from the patient section of our website https://www.snp.com.au/patients/collection-information/collection-notes/

Updates have been made to these notes:

- Ambulatory Blood Pressure Monitoring – item 35395
- Blood Tests – item 34168
- Glucose Tolerance Test – item 34159
- Urine Collection – item 34160
- Dexamethasone – item 34157
- Urine Cytology – item 50721
- Urine 5-HIAA - item 34038

Please be aware that significant changes have been made to:

- Fasting Blood Tests - item -34167 (there has been a change made to the number of days a patient must avoid alcohol)
- Seminal Collection - item 35170.

For more information, or a list of the latest notes and versions, please contact your Medical Liaison Manager (**P**: 1300 767 284 **E**: education@snp.com.au).

Familial hypercholesterolaemia testing

By A/**Prof Damon Bell** MB ChB PhD FRCPA FFSc FRACP

Genetic testing is the gold standard for diagnosing a patient with Familial Hypercholesterolaemia (FH). The development of Proprotein Convertase Subtilisin/Kexin type 9 (PCSK9) inhibitors has been shown to be very efficacious in patients with FH, reducing LDL-cholesterol by greater than 50% (combined with statins and ezetimibe). PCSK9 inhibitors are available on the PBS for patients meeting certain criteria.

Sullivan Nicolaides Pathology and Sonic Genetics have a comprehensive FH service to assist specialists and general practitioners identify patients with FH. The laboratory will highlight patients at risk of FH based on their LDL-c, and offer FH genetic testing with professional pre- and post-test genetic counselling. With consent from doctor and patient, Sonic Genetics can also assist in identifying family members for cascade testing.

Familial hypercholesterolaemia is a common inherited disorder of low density lipoprotein cholesterol (LDL-c) catabolism, which leads to increased lifetime LDL-c exposure and premature atherosclerotic cardiovascular disease (asCVD).

There is strong evidence that early treatment reduces the excess cardiovascular risk associated with this genetic condition.[1] Although we currently have very good methods to diagnose and treat people with FH, the largest challenge in FH management worldwide is to increase awareness and to detect people with FH.

FH occurs in 1:250 people, that is, there are ~100,000 people with FH in Australia. However, less than 10% are diagnosed, with ~90,000 remaining undiagnosed and under-treated. People with untreated FH have a risk of asCVD of ~50% in men by age 50 years, and of ~30% in women by age 60 years.[2] A healthy lifestyle, including low-fat diet and regular exercise, together with lipid lowering with statins and ezetimibe, are the cornerstones of therapy. Lipid lowering therapy dramatically reduces the risk of asCVD, and is currently recommended from the age of 10 years.[3] Despite these treatments, the desired LDL-c targets (LDL-c <2.6 mmol/L without asCVD, or <1.8 mmol/L with asCVD),[1] were achieved in only a minority of patients with formally diagnosed FH until the release of proprotein convertase subtilisin/kexin type 9 (PCSK9) inhibitors. These newer agents can reduce LDL-c by over 50%, in addition to the effect of statins and ezetimibe, and are available on the PBS for people with FH who meet LDL-c and clinical criteria.

Genetic testing is the gold standard for diagnosing patients with FH. The genetic diagnosis is important, as patients with genetically confirmed FH have a higher asCVD risk than non-FH patients for any given LDL-c. The relatives of a patient with a pathogenic variant are at risk of having inherited that variant and developing FH.[2] FH is inherited as an autosomal dominant trait, and genetic testing of at-risk relatives (cascade testing) is more accurate than biochemical testing, and has been demonstrated to be cost effective.[2] Genetic testing for the initial member of a kindred identified at risk of FH (index case), and family testing for the family's variant in first (50% risk) and second (25% risk) degree family members, is now rebated by Medicare from 1st May 2020.

A/Prof Damon Bell is a chemical pathologist and endocrinologist based in Perth WA with expertise in the diagnosis and management of inherited and acquired lipid disorders, including familial hypercholesterolaemia. His PhD focused on optimising the detection and management of familial hypercholesterolaemia, which remains one of his major research interests.

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References:

1. Pang J, Sullivan DR, Brett T, Kostner, KM, Hare DL, Watts GF. Familial hypercholesterolaemia in 2020: A leading tier 1 genomic application. Heart, Lung Circ 2019.doi.org/10.1016/j.hlc.2019.12.002
2. Lan NSR, Martin AC, Brett T, Watts GF, Bell DA. Improving the detection of familial hypercholesterolaemia. Pathol 2019;51(2):213-221
3. Luirink I, Wiegman A, Kusters D, Hof M, Groothoff J, de Groot E, et al. 20-Year follow-up of statins in children with familial hypercholesterolaemia. N Engl J Med 2019;381:1547–56.

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