AN ARTICLE FEATURING **Dr Charles Nelson**

The role of screening in familial cardiovascular disease

Good medical practice is to take a family history, including for familial cardiovascular disease, and, if indicated, screen your patient for the relevant disease. Conversely when familial cardiovascular disease is detected in your patient, ideally, the relatives should be screened.

It is my practice to provide my patient with a letter to pass on to their relatives alerting them and their general practitioner of the risk of the disease and role of screening. Adequately resourced specialised publicly funded clinics carry out "cascade screening": screening first-degree relatives (1stDR) and then screening the 1stDR of any relative in whom the disease was detected.

Familial hypercholesterolaemia has a prevalence of ~1 in 300, 18 times higher in the presence of atherosclerotic coronary disease (CAD). Diagnosis is via clinical scores. 1stDR should be screened with a clinical score. Inheritance is autosomal dominant.

Idiopathic dilated cardiomyopathy (IDC) has a prevalence of ~ 1 in 250. Up to over 50% of relatives of patients initially diagnosed with IDC are found to have dilated cardiomyopathy. 1stDR should be screened with echocardiogram and ECG and even if negative should be followed-up every 3-5 years as the phenotype may manifest in subsequent years. Ideally the proband should undergo genetic testing. 1stDR may then be screened for the causative gene with those positive being followed up. Most inheritance is autosomal dominant, though other patterns occurr.

Prevalence of hypertrophic cardiomyopathy (HCM) is 1 in 500. Probands should undergo genetic testing. 1stDR of probands with pathogenic genetic mutations should be screened with ECG and echocardiogram. If clinical evidence of HCM is detected genetic testing is optional. If no clinical evidence of HCM is detected genetic testing is recommended with positive cases being followed up and negative cases reassured.

Prevalence of hypertension is up to 47%. Ideally blood pressure should be assessed at least annually in every adult from age 18. Heritability of blood pressure is up to 50%. Hypertension is 2.5 times more likely in patients who have 2 hypertensive parents.

The 2019 European Society of cardiology guidelines state that screening for CAD "may be considered" in selected but asymptomatic high risk patients with diabetes e.g. those with atherosclerotic disease in non-coronary territory, high calcium score or proteinuria. ~30% of diabetic patients with no clinical evidence of coronary disease have obstructive CAD. The main change to management resulting from diagnosing obstructive CAD in an asymptomatic diabetic is prescription of low-dose aspirin.

Patients with a family history of sudden cardiac arrest (SCA) have a modest 1.5–1.8 fold increase in the risk of SCA. Relatives of patients with SCA should be screened for coronary risk factors along standard guidelines. Routine additional testing is not recommended. There is no evidence such testing identifies patient is at higher risk.

Up to 8% of males > 50yo have abdominal aortic aneurysm (AAA). Aneurysmal disease is the cause of death in 0.13% of males and 0.07% of females. A positive family history doubles the risk of AAA. It is recommended to screen with ultrasound males age 65-75 who have a first-degree relative with AAA or males who have ever smoked. Screening reduces mortality. Screening of females is not routinely recommended but may be reasonable in individual cases.

Thoracic aortic aneurysms (TAA) account for one third of hospital aortic aneurysm admissions (with the remainder being due to AAA's). 1stDR of patients with TAA should undergo screening with Echocardiography or CT. There is a 20% chance that a 1stDR will have TAA. For patients with a positive family history genetic testing is recommended. For relatives with positive genetic testing imaging with echocardiography or CT is recommended. For those gene positive but imaging negative, follow-up is indicated.

AAA is present in ~20% of patients with TAA. Up to 40% of patients with AAA have at least 1 iliac artery aneurysm (IAA). 86% of patients with IAA have AAA.

Patients with a popliteal artery aneurysm have a high incidence of aneurysm in the contralateral popliteal artery, femoral arteries and abdominal aorta and should be screened for these with ultrasound.

Bicuspid aortic valve (BAV) effects 1% of the population. The prevalence of bicuspid valve in first-degree relatives is ~10%. Inheritance is consistent with an autosomal dominant pattern with incomplete penetrance. Aortic dilatation is present in up to 84% of BAV patients and coarctation in 6%. 1stDR of BAV patients should be screened for BAV and thoracic aortic disease with echocardiography +/- MRI/CT.

The prevalence of congenital long QT syndrome (LQTS) is probably 1 in 1000. There are autosomal dominant and autosomal recessive forms. The prevalence of Brugada Syndrome is probably 1 in 1000. Inheritance is autosomal dominant with variable penetrance. 1st DR of patients with LQTS or Brugada Syndrome should be screened with ECG and genetic testing.

Thus screening plays a significant role in the care of patients and families with familial cardiovascular disease.

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