Case reports

A family with heritable hypertriglyceridaemia

A 44-year-old man with severe pancreatitis was diagnosed with type I hyperlipidaemia (hereditary chylomicronaemia) after poorly responsive hyperlipidaemia and attacks of pancreatitis. He recovered after assisted ventilation and drainage of a pseudocyst. He had a dramatic improvement of plasma lipids after withdrawing drugs and instituting a low-fat diet. A simple assay of lipolysis revealed no post-heparin lipolytic activity.

His sister had presented to the clinic previously with recurrent pancreatitis ascribed to type V hyperlipidaemia. She responded well to dietary modification and a fibrate, but was prone to develop hypertriglyceridaemia and pancreatitis after dietary indiscretion. She developed diabetes mellitus but had an uncomplicated pregnancy on a very low-fat diet and insulin treatment. Investigation of the family members revealed other patterns of dyslipidaemia, including Fredrickson IIa and IIb.

The lipolytic assay showed that the diabetic mother had normal post-heparin lipase activity but the sister had partial lipase activity. Since lipolysis was not reconstituted with pre-heparin serum from a normal person, apoCii deficiency was excluded. Investigations by polymerase chain reaction excluded the 2 commonest mutations found with type I hyperlipidaemia in our clinic, but the investigation is incomplete.

Partial lipoprotein lipase deficiency is associated with various lipoprotein phenotypes, depending on additional environmental and genetic factors. Hereditary chylomicronaemia should be considered in patients with severe hypertriglyceridaemia, especially if they respond poorly to lipid-modifying drugs. A very low-fat diet can control the dyslipidaemia and reduce the risk of pancreatitis.

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Mitral stenosis: often missed and potentially lethal

Mitral stenosis (MS) is often missed but potentially lethal.

A 41-year-old woman presented with severe dyspnoea. She had suffered a cerebrovascular accident at 35 years of age. She was in rapid atrial fibrillation (AF), and an embolic left femoral artery occlusion resulted in amputation. Acute myocardial infarction complicated her course. Clinical features were those of MS confirmed echocardiographically. The severe embolic complications (peripheral arterial, coronary and cerebrovascular) prompted referral for mitral valve replacement surgery as left atrial thrombus precluded percutaneous valvuloplasty.

Another patient, a 43-year-old woman, had increasing dyspnoea and wheeze, asthma and 'mild' MS on echocardiography. On presentation the differential diagnosis was between severe asthma with AF and MS.

Echocardiography demonstrated severe MS and impaired left ventricular function attributed to tachycardia-mediated cardiomyopathy. Rate control was achieved with verapamil, digoxin and a β-blocker tolerated despite the history of 'asthma'. Severe MS was confirmed at catheterisation and was successfully corrected surgically.

These cases illustrate several clinical issues:

- MS, the sequela of rheumatic fever, is all too common in South Africa.
- It may cause, as in these young women, devastating illness, prolonged hospitalisation, loss of a limb, myocardial infarction or high-risk surgery.
- The clinical manifestations are protean. Thromboembolic complications may be the first manifestation in 25% of patients. Haemoptysis and chest pain, rather than dyspnoea, may be presenting symptoms.
- Symptoms may be incorrectly attributed to co-existing conditions.

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single suture

Tube feeding in dementia

The practice of tube feeding elderly, demented patients does not prolong or improve the quality of life and can, in some cases, actually shorten life. This is the conclusion reached by John Hoffer in a discussion article in a recent *British Medical Journal*. We need to remember that elderly people who eat very little are unlikely to be starving. They simply have very low energy requirements and are probably in metabolic homeostasis. Good practice, according to Hoffer, comprises regularly weighing patients to make sure that weight loss is not life-threatening and paying attention to the quality, characteristics and presentation of food.

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