Screening & Management of Hypercholesterolaemia in Childhood

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Children with familial hypercholesterolaemia are at high risk of developing coronary artery disease (CAD) in early adulthood, and should be identified in childhood.

The gene frequency of the heterozygous form of this autosomal dominant condition is about 1 in 500 in the UK population. Other inherited disorders of lipoprotein metabolism are rarely expressed in childhood.

SCREENING STRATEGIES

Population screening for hyperlipidaemia in children is **not recommended**. It identifies large numbers of children with polygenic hypercholesterolaemia of uncertain prognostic significance and small numbers with familial hypercholesterolaemia.

Case finding by **family tracing** is an effective strategy for identifying most children with familial hypercholesterolaemia. Screening should be based on:

- a family history of familial hypercholesterolaemia or
- premature CAD (angina or myocardial infarction).

The age cut-off chosen to define premature CAD is to some extent arbitary. Occurrence in men before 50 years and women before 55 years is more specific but less sensitive than corresponding ages of 55 and 60 years.

Age of testing. Children should be tested before the age of 10 years but usually not before the age of two years. The age chosen should take into account the onset of CAD within the family and the wishes of the parents.

Blood samples. Non-fasting capillary or venous samples can be used for screening measurement of total cholesterol. If the concentration is above 5.5mmol/l, which is the 95th centile in childhood, a fasting venous blood sample should be taken for measurement of total and high-density lipoprotein (HDL) cholesterol and triglyceride. The low density lipoprotein (LDL) cholesterol can then be calculated (see appendix).

DIAGNOSTIC CRITERIA

Diagnosis of familial hypercholesterolaemia in a child under 16 years should be based on finding:

- total cholesterol concentration above 6.7 mmol/l
- together with an LDL cholesterol concentration above 4.0 mmol/l
- requires at least two measurements to be made more than one month apart.

MANAGEMENT

Clinical care. Affected and possibly affected children should be referred for specialist hospital follow-up.

Clinical management includes:

- Avoidance of cigarette smoking
- Encouragement of physical exercise
- Dietary advice to reduce total fat intake to 30-35% of total calories. Dietary energy must be sufficient to support normal growth, but saturated fatty acid intake should account for no more than 10% of total calories. An experienced dietician should provide advice for the family.

Drug therapy. Lipid-lowering drug therapy should only be prescribed by a specialist clinic. It should usually be restricted to children aged 10 years or more, dependant on the age of onset of CAD in the family. It is less often indicated in girls than in boys since coronary risk in early adulthood is substantially lower in women. Parents should be aware of the limitatioins of the short-term efficacy and safety data for drug use in childhood, but should recognise that drug treatment will almostcertainly be required later in adulthood.

Appendix

LDL cholesterol can be calculated using the Friedewald formula below provided the triglyceride concentration is less than 4.5 mmol/l :

LDL Cholesterol = total cholesterol - HDL cholesterol - (triglycerides/2.19mmol/l)

Further Reading:

Hyperlipidaemia in childhood. Neil A, Rees A, Taylor C, eds Royal College of Physicians, London, 1996.

Wray R, Neil HAW, Rees JAE. Screening for hyperlipidaemia in childhood:Recommendations of the British Hyperlipidaemia Association. J R Coll Physicians (Lond) 1996;30:115- Goldstein JL, Hobbs HH, Brown MS. Familial Hypercholesterolaemia. In : The Metabolic and Molecular Basis of Inherited Disease *Scriver CR, Beaudet AL. Sly WS. Valle D. eds) 7th edition. McGraw Hill, New York 1995, pp 1981 - 2030

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