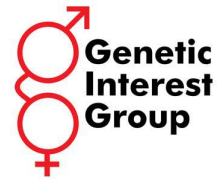


NICE consultation on Familial Hypercholesterolaemia: the identification and management of patients with familial hypercholesterolaemia.



A response from the Genetic Interest Group

The Genetic Interest Group (GIG) is a UK-wide alliance of organisations with a membership of over 130 charities which support children, families and individuals affected by genetic disorders. Its primary goal is to promote awareness and understanding of genetic disorders so that high quality services for people affected by genetic conditions are developed and made available to all who need them. GIG is the only UK organisation of its kind.

The Genetic Interest Group welcomes the opportunity to comment on this Draft Scope consultation, and would be happy to discuss any of this response further.

Homozygous Hypercholesterolaemia

The Genetic Interest Group endorses all procedures to treat homozygous hypercholesterolaemia. Early identification of the condition and quick treatment is paramount for a condition with early onset and such serious symptoms. Therefore pre-natal diagnosis of those families at risk of having homozygous FH children is an important and necessary measure.

Information, genetic counselling, and lifestyle advice

The Genetic Interest Group has much experience of the value of good quality information to educate patients and their families about the condition for which they have received a diagnosis. Information such as this helps the patient understand and plan for the future, and can assuage feelings of guilt within the family.

Genetic Counselling is important for patients and their families to understand the implications of an inheritable disorder; it can also be a useful opportunity for patients to ask any other questions they may have about the condition.

Screening

Cascade screening depends on accurate and comprehensive identification of index cases. Rare conditions such as these are more economically treated with targeted screening of groups at elevated risk; the Genetic Interest Group supports this choice of screening strategy.

Quicker diagnoses will lead to earlier decisions on management of the condition, and a better patient experience. Quick diagnoses are particularly important for homozygous hypercholesterolaemia.

The Genetic Interest Group asserts that genetic testing should be used in hypercholesterolaemia screening as and when it can be proven that genetic testing confers an advantage in diagnosis over other methods available.

Conclusion

The Genetic Interest Group endorses the aims of this consultation: to provide a NICE guideline for treating patients with either heterozygous hypercholesterolaemia, or homozygous hypercholesterolaemia. It is important to change the profile of heterozygous hypercholesterolaemia from a condition diagnosed after a coronary heart disease episode, to a risk factor that is identified early and treated successfully.

A handwritten signature in black ink that reads "Alastair Kent".

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