Direct to consumer genetic tests - haemochromatosis

There are a number of commercial services currently providing direct-to-consumer genetic tests in the United Kingdom. These range from broad genome analysis (e.g. 23andMe) to others providing testing for specific pathogenic variants associated with disease prediction (e.g. *HFE* testing offered by Haemochromatosis UK). There are no existing service standards for such services nor any formal regulatory body/framework.

We are aware that some members of the public receiving *HFE* genetic results have received misleading clinical information regarding the risk of haemochromatosis which has caused confusion and anxiety to individuals as well as an additional burden to NHS healthcare providers. **A diagnosis of haemochromatosis cannot be based on genotypic information alone**. Iron studies and a clinical assessment are always needed before a diagnosis can be made. Even with the highest risk genotype (pC282Y homozygosity) a significant number of individuals do not get significant iron overload. A false diagnosis of haemochromatosis based on genotypic information alone might be harmful by distracting both the patient and their clinician from other possible causes for a patient's symptoms.

We recommend that for patients referred with a direct-to-consumer genetic test result, the reported pathogenic variant is verified by a repeat test undertaken in a CPS accredited diagnostic laboratory. Each patient requires individual assessment of disease risk - irrespective of any inference provided by the commercial test provider.

Evidence-based information regarding haemochromatosis disease risk associated with specific *HFE* genotype based on UK Biobank data is available via the Exeter University, Epidemiology and Public Health Group (https://sites.exeter.ac.uk/ironoverload/). Additional clinical support can be sought through the BASL/BSG/BSH haemochromatosis special interest group (SIG) via their host organisation The British Association for the Study of the Liver (BASL).

We fully encourage all commercial providers of genetic testing and patient representative organisations to work closely with expert clinical groups in the development of appropriately worded reports to accompany test results.

Dr Jeremy Shearman BSG, BASL
Dr Bill Griffiths BASL
Dr Charlotte Bradbury BSH
On behalf of the BASL/BSG Haemochromatosis Special Interest Group