



Guidelines for Hereditary Pancreatitis Testing

To Test or Not To Test

The following criteria for hereditary pancreatitis (HP) genetic testing are taken from:

Ian Ellis, Markus M. Lerch and David C. Whitcomb. Genetic Testing for Hereditary Pancreatitis: Guidelines for Indications, Counselling, Consent and Privacy Issues. *Pancreatology* 2001; 1:405-415.

We are aware that with the introduction of any new test there is a desire to 'try it out'. We set out specific indications for the decision to offer diagnostic molecular genetic testing for HP. Outside of defined Ethics Committee Approved Research Protocols, the indication for PRSS1 mutation analysis in a symptomatic patient should be any of the following:

- 1) *Recurrent (2 or more separate, documented episodes with hyper-amylasaemia) attacks of acute pancreatitis for which there is no explanation (anatomical anomalies, ampullary or main pancreatic strictures, trauma, viral infection, gallstones, alcohol, drugs, hyperlipidaemia, etc.), or*
- 2) *unexplained (idiopathic) chronic pancreatitis, or*
- 3) *a family history of pancreatitis in a first-degree (parent, sib, child) or second degree (aunt, uncle, grandparent) relative, or*
- 4) *an unexplained episode of documented pancreatitis occurring in a child that has required hospitalization, and where there is significant concern that HP should be excluded (see 'The Genetic Testing of Children' below), or*
- 5) *patients with pancreatitis eligible for an Ethics Committee Approved Research Protocol.*