Juvenile polyposis is caused by germline mutations in *SMAD4* or *BMPR1A,* but there remains a set of JPS families and sporadic cases without detectable mutations in these genes. We wish to ascertain JPS cases of unknown genetic origin to explore the possibility that undiscovered JPS gene(s) exist.

Cases or families would be eligible if any affected member capable of providing blood or DNA has had 2+ juvenile polyps at any age. Other unusual features that are part of the JPS syndrome (e.g. AV malformations) should be noted, but these do not form part of the inclusion criteria.

Ideally, mutations in *SMAD4* or *BMPR1A* would have been excluded. Mutations in other TGFβ/BMP pathway genes need not have been excluded. We propose to search for new JPS genes using sequencing, supplemented where possible by other techniques including linkage analysis and studies of polyps or colorectal cancers.

All work will be performed as a collaboration within the European Hereditary Tumour Group.

Andrew Latchford
Karl Heinimann
Ian Tomlinson