The UKGTN evidence provided in this response is outlined in relation to medical research investigating the genetic cause of human disease which will ultimately lead to the provision of molecular/cytogenetic diagnostic testing through clinical service.

1. The UKGTN propose a mandatory requirement for research organisations to seek patient opinion through full consultation with patient representative groups. This partnership would facilitate the communication of research outcomes to patients as part of the patient follow-up programme, retain patients as willing participants and recruit their relatives to the research study.

2. The UKGTN emphasises the need for international collaboration to strengthen the evidence base currently lacking for very rare genetic disorders. This would facilitate closure of the gap in clinical service provision for these tests.

3. In line with DH guidance, the UKGTN propose a mandatory requirement for all key participants including lead commissioners to provide letters of support at the time of grant application. This should include large multi-centered studies. As a consequence, PCTs would need to be part of the decision making process. It has been highlighted that not all PCTs/SHAs are familiar with the guidance concerning excess treatment costs and would seek guidance covering these issues.

4. The UKGTN propose where genetic testing of patient samples is required as part of a research study, Principal Investigators should seek the collaboration of an accredited diagnostic laboratory to perform these tests where possible and include funding for such testing in the grant application. This ensures that patients receive quality controlled results from the testing and avoids the need for them to be repeated later by a diagnostic lab before being passed on to the patient.