



## Checklist for diagnostic genetic testing

### Unexplained physical symptoms identified

- Obtain a detailed medical family history and record findings using a three-generation genetic pedigree
- Identify the right test for your patient using the National Genomic Test Directory or equivalent directory for your nation  
*Use the PanelApp to find out more about the genes being tested*
- Confirm eligibility of your patient using the relevant test directory for your nation
- Obtain informed consent: Do they understand the implications of the test for themselves and the family?
- Document informed consent: Write down your discussions in the patient notes or using a Record of Discussion (RoD) form  
*If testing using whole genome sequencing (WGS), completing the RoD is mandatory*
- Consider contacting your local clinical genetics service for advice and support before the test

### Arranging a genetic test

- Complete the Genetic Testing Request Form. These will vary by trust and are available on your local Genomic Laboratory Hub website
- Send to your local Genomic Laboratory Hub, unless otherwise specified

### Interpreting the results

- Clearly and empathetically communicate test results to the patient
- Signpost to relevant support resources
- Consider referral to a clinical geneticist and/or genetic counsellor, depending on test result
- Refer to nominated ultra-specialist centre for further support and access to disease-specific therapies**