

WGS : 7 Steps for Success



- 1** Arrange a **trio** for paediatric cases to help actionable results.
Without DNA from both parents, scientists cannot fully analyse any rare variants that are detected
- 2** Store the patient **sample correctly** (eg blood samples in purple top EDTA).
DNA will be damaged if it's not in the correct tube. Formalin will damage tissue DNA too.
- 3** Complete all forms **electronically**. Handwritten forms will be rejected.
- 4** Include the patient's **ethnicity** to improve accuracy of results.
We need to compare results with population specific data to understand if variant could be causative
- 5** Explain why your patients meet the eligibility criteria & include ALL **HPO** terms (Human Phenotype Ontology). Even those that might not seem relevant, otherwise we might miss relevant findings. Find HPO terms on the HPO website & select terms that start with HPO.
- 6** Email the **Record of Discussion** & Test Order Form to gstt.wgs@nhs.net
We can't start testing until we have ALL forms for family members being tested.
One Record of Discussion per person. One Test Order Form per trio.
- 7** Look out for an **email** from the WGS team.
We'll email you if there is a problem. Testing will be cancelled after 16 weeks if the referral isn't complete.



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