

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 the relevant regional laboratory.

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 duo or 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.