Fetal Medicine:
Pathway and checklist for R21 testing ongoing pregnancy

NHS Central and South Genomics

1 Fetal anomalies identified

Fetal anomalies identified on ultrasound scan consistent with R21 inclusion criteria.

- [ ] Contact on-call clinical geneticist to discuss findings.
- [ ] Email R21 request with full clinical information to:
  Home LGL/GLH:
  Testing GLH: **bwc.rglprenatalexome@nhs.net**
  Clinical Genetics: West Midlands: **bwc.prenatalclinical@nhs.net** | Wessex: **geneticsprenatal@uhs.nhs.uk** | Oxford: All cases managed via clinical genetics

LGL: Local Genetics Laboratory GLH: Genomics Laboratory Hub

2 Consent & Record of discussion

- [ ] Provide parents with R21 information leaflet.
- [ ] Consent: Complete a 'record of discussion form (RoD)' for each parent being tested. The Testing GLH should be informed that consent has been provided. The RoD can be stored in the patient maternity notes*.

*Can be after fetal sample has been sent and/or before R21 approval has been obtained.

3 Send samples

Send fetal sample and maternal and paternal bloods to Home LGL/GLH. State if R21 approved or awaiting approval.

- [ ] Include: 'Rare and Inherited Disease Referral' form for each sample (ensure the paternal sample form refers to the maternal/prenatal sample), scan reports and growth charts/other relevant tests if available.
- [ ] **Mark all samples as URGENT and for R21 testing.**

R21 testing is run once a week. Establish sample receipt requirements with Home GLH and consider timelines for sample collection and transport.

4 Liaise with Testing GLH

- [ ] **Email 'R21 request' form to the Home LGL/GLH and Testing GLH.**

The home LGL/GLH will add the results and sample information and email the updated form to the Testing GLH (Birmingham Women's & Children's Trust).

5 Signpost to support

- [ ] Inform parents of turnaround time of 2 - 3 weeks.
- [ ] **Signpost to support resources.**

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