NHS
North East and Yorkshire Genomics

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5th October 2026

Dear Colleague,

Change to follow up testing of parental samples for R22 and R318 referrals

Your routinely send us material for genetic testing following pregnancy loss. The two main clinical indications for referral are:

R22 Fetus with a likely chromosomal abnormality (for non- recurrent pregnancy losses)

R318 Recurrent miscarriage with products of conception available for testing

We routinely test by SNP array. Currently where there is an abnormal or uncertain result, we may follow up by testing parents either by a targeted karyotype or SNP array. This is indicated on the report.

From 26th October 2026 we will be modifying this practice slightly to align with pending best practise guideline changes and with our partner laboratories (Sheffield and Newcastle) in the NEYGMS. This only affects those reports where a chromosomal trisomy is detected:

- For apparent chromosomal trisomy, which is a relatively common finding in pregnancy loss, we will only follow up parents for further testing where the chromosomes involved are the acrocentric chromosomes: 13,14,15, 21 and 22. For these chromosomes, there is a small but raised risk that malsegregation of a balanced structural rearrangement (typically a Robertsonian translocation) carried by a parent has resulted in the trisomy in this case, and there is a slightly increased risk of recurrence related to this, in any future pregnancy. We will request parental samples for follow up targeted testing.
- The risks, following pure trisomy for any other chromosome are very low, both for a parental structural rearrangement and consequently for any recurrence risk related to a structural re-arrangement, and we will now not request parental samples for follow up.
- For any other finding on the initial SNP array analysis, we will continue to follow up as appropriate, which will be clearly indicated on the report.

If you have any queries regarding this change, please contact myself james.steer@nhs.net or andrea.coates1@nhs.net

Yours Faithfully,

James Steer
Head of Laboratory