Parental cytogenetic testing

Introduction

Parental cytogenetic testing may be requested following the conception of a cytogenetically abnormal fetus, the birth of a cytogenetically abnormal child, the loss of a pregnancy with a suspected chromosome abnormality, or an unexplained stillbirth/neonatal death.

Referrals

Samples can be referred for a wide variety of reasons (see below):

- parental karyotyping after pregnancy loss of a fetus <24 weeks with multiple congenital abnormalities or severe IUGR, or unexplained stillbirth/neonatal death >24 weeks
- parental testing following MLPA/QF-PCR testing of products of conception if an unbalanced structural chromosomal abnormality is identified or a result is not obtained for technical reasons i.e. a sample was received but failed to produce a result
- parental testing after three or more unexplained miscarriages if the patient is from a hospital with a Service Level Agreement (e.g. Chase Farm, Barnet, Whittington and Royal Free Hospitals); this testing can also be performed on a private patient basis for patients from other hospitals
- parental testing if there have been two identical aneuploidies suggesting parental mosaicism
- parental testing following an abnormal cytogenetic result in a fetus or child NB the proband’s report will state if parental testing is indicated

Please note that parental testing after three or more unexplained miscarriages is not routinely offered (in line with the 2011 RCOG ‘green top guidelines’).

Service offered

The format of the parental testing will vary depending on the abnormality suspected or detected in the proband. The testing may be performed by karyotyping (full or targeted), fluorescent in situ hybridisation (FISH), qPCR, QF-PCR, MLPA, or rarely microarray. More than one technique may be used.

Technical

If the test is karyotyping or FISH, blood cultures are grown and harvested to yield metaphase cells which are analysed using light microscopy. Extracted DNA is used for qPCR, QF-PCR, MLPA and microarray testing.

Targeted analysis is performed in the following circumstances:-

- Parental follow-up of copy number changes detected in products of conception following pregnancy miscarriage
- Known abnormality or familial structural abnormality (e.g. family history of a known translocation, child has chromosome abnormality) – targeted analysis: If a patient requires testing because of a family history of a structural chromosome abnormality (e.g. translocation, inversion) and a written copy of the abnormal family member’s cytogenetic result including ISCN is available

Target reporting time

Routine analysis – 28 days

If clinical need indicates that an urgent result is required (e.g. current ongoing pregnancy) – 10 days.