**Fact Sheet for Requesting Genetic Tests on Feto-Maternal Samples**

**There are 3 genetic testing request forms**

1. **The prenatal request form for fetal samples and maternal samples for NIPD**
2. **The rare disease request form for neonatal and all postnatal samples**
3. **An additional rapid fetal exome request form for indication R21**

**\*A separate maternal sample to exclude maternal cell contamination (MCC) is required for all invasive procedures including amnio, CVS, fetal blood, cord blood and products of conception unless maternal DNA is already stored in the lab. Please indicate on the pre-natal form whether a maternal sample is accompanying the pre-natal sample**

**The prenatal request form has the maternal identifiers on it. The rare disease form has the neonates details on it**

**With the advent of the National Genomic Test Directory the request form requires an R number and a test description. For Fetal testing these are as follows:**

**R401 Common aneuploidy testing - prenatal (e.g. QFPCR)**

**R318 Recurrent miscarriage with products of conception available for testing (QFPCR and microarray). These would usually be first trimester miscarriages. It may be more appropriate to request R28 microarray or R14 neonatal exome for later pregnancy loss R22 Fetus with a likely chromosomal abnormality (QFPCR and microarray)**

**R21 Fetal anomalies with a likely genetic cause – rapid fetal exome (after QFPCR) – please note this has to involve Clinical Genetics and requires an additional form**

**R240 diagnostic testing for known familial mutation**

**R321 maternal cell contamination testing – if coming in separate to the fetal sample**

**R320 invasive prenatal diagnosis requiring fetal sexing**

**R251 Non-invasive prenatal sexing**

**R249 NIPD using paternal exclusion testing for very rare conditions where familial mutation is known . This can only be requested by Clinical Genetics**

**R250 NIPD for congenital adrenal hyperplasia - CYP21A2 haplotype testing**

**R304 NIPD for cystic fibrosis - haplotype testing**

**R305 NIPD for cystic fibrosis - mutation testing**

**R306 NIPD for Apert syndrome - mutation testing**

**R307 NIPD for Crouzon syndrome with acanthosis nigricans - mutation testing**

**R308 NIPD for FGFR2-related craniosynostosis syndromes - mutation testing**

**R309 NIPD for FGFR3-related skeletal dysplasias - mutation testing**

**R310 NIPD for Duchenne and Becker muscular dystrophy - haplotype testing**

**R311 NIPD for spinal muscular atrophy - mutation testing**

**R389 NIPD - pre-pregnancy test work-up. This needs to be requested on the rare disease request form**

**R92 Thalassaemia and other hamoglobiopathies**

**R94 Sickle Cell anaemia**

**R184 Cystic Fibrosis diagnostic test**

**The testing criteria can be found in detail at** <https://www.england.nhs.uk/publication/national-genomic-test-directories/>

**If the test you require is not on this list please discuss with Clinical Genetics and the laboratory.**