Noninvasie Prenatal Diagnosis (NIPD) for single-gene disorders

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We offer an ISO 15189:2012 accredited service for non-invasive prenatal diagnosis (NIPD) for single-gene disorders at our National Health Service (NHS) Regional Genetics laboratory based at Great Ormond Street Hospital, London. Since 2013 we have offered a United Kingdom Genetic Testing Network (UKGTN) approved service for FGFR3-related skeletal disorders, including achondroplasia and thanatophoric dysplasia. We also have UKGTN approval for FGFR2-related craniosynostosis, including Apert and Crouzon syndrome, and for paternal mutation exclusion for cystic fibrosis. We have expanded our service to improve availability of NIPD for couples at risk of single-gene disorder and to offer definitive diagnosis for cystic fibrosis.