Uniparental Disomy of Chromosome 16

INTRODUCTION
Uniparental disomy (UPD) is the abnormal situation in which both copies of a chromosome pair are inherited from only one parent, with no copies inherited from the other parent. UPD has been observed to result through several mechanisms including trisomy rescue and gametic complementation, and from the presence of structurally abnormal chromosomes, including Robertsonian translocations.

Most cases of uniparental disomy involving chromosome 16 (UPD16) arise from an initial trisomy and are maternal in origin. The prominent phenotype of matUPD16 is intrauterine growth retardation. Growth may normalize postnatally and other features may include anal atresia, heart defects, inguinal hernia, clubfoot, hypospadias, elbow dislocation, lung cysts and thoracic skin tags, pulmonary hypoplasia, hypothyroidism and absence of one kidney, although these features may be partly due to hidden mosaic trisomy 16. Mental development ranges from normal to severely delayed.

REFERRALS: Only accepted from Clinical Genetics

TESTING
Diagnostic testing:
- Clinically affected individuals
- Proband at risk of having UPD16 following prenatal detection of placental trisomy 16

Prenatal testing, where possible arranged in advance:
- Fetus is shown to have a chromosome 16 derived marker chromosome or mosaicism for trisomy 16 and is therefore at risk of UPD16

STRATEGY AND TECHNICAL INFORMATION
- Microsatellite marker analysis (parental samples are required and will be requested prior to analysis)
- The sensitivity of this test is unknown, but will be dependent on the availability of family members, the informativity of individual families and the location of informative markers, in cases of segmental UPD.

TARGET REPORTING TIMES
Diagnostic testing: 10 days
Prenatal testing (includes maternal contamination check): 3 days

N.B. Details are correct for the date of printing only - Last updated 02/09/2015