Thrombocytopenia-Absent Radius Syndrome (TAR)

Clinical Background and Genetics

- Thrombocytopenia-absent radius (TAR) syndrome is characterised by hypomegakaryocytic thrombocytopenia and bilateral radial aplasia in the presence of both thumbs.
- These characteristic patterns differentiate TAR syndrome from other conditions with involvement of the radius, namely Holt-Oram syndrome, Roberts syndrome and Fanconi Anaemia in which the thumb is usually absent or severely hypoplastic.
- Additional skeletal features associated with TAR syndrome include shortening and, less commonly, aplasia of the ulna and/or humerus.
- The hands may show limited extension of the fingers, radial deviation and hypoplasia of the carpal and phalangeal bones.
- The lower limbs are frequently involved but to a lesser extent than the upper limbs.
- Dislocation of the hips and subluxation of the knees resulting in coxa vara are common.
- Other frequent associations are congenital heart disease and a high incidence of cow's milk intolerance.
- The genetic basis of TAR syndrome remains unclear; current understanding is that TAR syndrome is associated with a microdeletion on 1q21.1 that is necessary but not sufficient to cause the phenotype.
- The inheritance pattern in TAR syndrome is complex, it is thought that the presence of an unknown modifier (mTAR) is required to cause the phenotype.
- Diagnostic testing is based on testing for the common deletion.

Level 1

- Test for deletion of the TAR critical region at 1q21.1 using MLPA.

Target reporting Time and Indicative Cost

- MLPA 10 days

Please contact the laboratory for up to date prices.

Referrals

- Given the incomplete understanding of TAR Syndrome inheritance only diagnostic cases are accepted for testing.
- Confirming the diagnosis of TAR syndrome relies on identifying the common deletion.
- Referrals are only accepted from Clinical Geneticists.

Clinical Advice

If clinical discussion is required we would recommend contact with Dr Ruth Newbury-Ecob, Clinical Genetics, St. Michael’s Hospital, Bristol (Tel: 0117 342 5652).