Diagnosis of Smith-Magenis Syndrome

Diagnosis of Smith-Magenis syndrome is confirmed through genetic testing after initial clinical findings (i.e. physical characteristics).

Cytogenetic testing (analysis of the number and structure of chromosomes) is used to detect the deletion and molecular genetic testing is used to determine whether there is a deletion on chromosome 17p11.2 including RAI1 or a mutation of the RAI1 gene.

In the clip below, parents give some advice about having a child with Smith-Magenis syndrome

If you have just received a diagnosis of SMS for your child, you may find this information booklet useful. More information about the diagnosis procedure can be found on the GeneReviews website. (Please note: this material is aimed at clinicians and researchers)