Genetics of Smith-Magenis Syndrome

After Smith-Magenis syndrome was first described it was subsequently associated with a de novo deletion on chromosome 17p11.2. A de novo deletion is a ‘new’ deletion that was not inherited from either parent.

More recently, individuals demonstrating features of Smith-Magenis syndrome caused by a mutation of the retinoic acid-induced 1 (RAI1) gene located on chromosome 17p11.2 have been described, leading researchers to suggest that this gene is implicated in most of the features of the syndrome. The mutation of gene RAI1 is responsible for less than 10% of cases.

Visit the Smith-Magenis Syndrome Foundation page to read more about genetics.