Mowat-Wilson Syndrome

National Cancer Institute

Source

National Cancer Institute. Mowat-Wilson Syndrome. NCI Thesaurus. Code C74999.

A rare autosomal dominant syndrome caused by mutations in the ZEB2 gene. It is characterized by mental retardation, and a distinctive facial appearance (wide set eyes, uplifted earlobes, broad nasal bridge, prominent chin, and a smiling expression). The majority of patients have Hirschsprung disease (colonic enlargement and constipation due to intestinal blockage).