Mowat-Wilson Syndrome

Alternative Names
Microcephaly, Mental Retardation, and Distinct Facial Features, with or without Hirschsprung Disease
Hirschsprung Disease Syndrome
Hirschsprung Disease-Mental Retardation Syndrome
Goldberg-Shprintzen Syndrome

WHO International Classification of Diseases
Congenital malformations, deformations and chromosomal abnormalities

OMIM Number
235730

Mode of Inheritance
Autosomal recessive

Gene Map Locus
2q22

Description
Mowat-Wilson syndrome is a multiple congenital anomaly syndrome. All patients have typical dysmorphic features in association with severe intellectual disability, and nearly all have microcephaly and seizures. Congenital anomalies, including Hirschsprung disease (HSCR), congenital heart disease, hypospadias, genitourinary anomalies, agenesis of the corpus callosum, and short stature are common.

Molecular Genetics
Mowat-Wilson syndrome results from sporadic heterozygous deletions or truncating mutations of the ZFHX1B (SIP1) gene on chromosome 2q22. The developmental ZFHX1B expression pattern fully explains the clinical spectrum observed in patients with Mowat-Wilson syndrome by haploinsufficiency of this gene.

Epidemiology in the Arab World

Morocco
In 1998, Brooks et al. reported three children from a large, consanguineous, Moroccan family with Hirschsprung disease, mental retardation, microcephaly, and specific craniofacial dysmorphism. The association of these abnormalities in these children represented the Goldberg-Shprintzen syndrome (Mowat-Wilson syndrome).

United Arab Emirates
Sztriha et al. (2003) reported a girl who had Hirschsprung disease in association with distinct facial appearance, microcephaly, agenesis of the corpus callosum and mental retardation (Mowat-Wilson syndrome).

References

Contributors
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