Martsolf Syndrome

**Alternative Names**
- Cataract-Mental Retardation-Hypogonadism
- Microcephaly-Mental Retardation-Cataract-Hypogonadism Syndrome

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

**OMIM Number**
212720

**Mode of Inheritance**
Autosomal recessive

**Gene Map Locus**
1q41

**Description**
Martsolf syndrome is a rare genetic disorder that is characterized by the association of severe mental retardation, cataract, and hypogonadism. Other anomalies presented in some patients with Martsolf syndrome include short stature, “old looking” face, mottled retina, minor digital abnormalities, microcephaly, narrow hands and feet, hypertelorism, cardiomyopathy, and cardiac failure. This condition is more prevalent in Jewish than other ethnic groups.

**Molecular Genetics**
Martsolf syndrome is found to be familial. In three reported families, the parents of the affected sibs were consanguineous, which indicates an autosomal recessive mode of inheritance. Some families have shown mutations in the RAB3 GTPase activating protein subunit 2 (non-catalytic) [RAB3GAP2] gene, which is thought to have an important role in neurodevelopment.

**Epidemiology in the Arab World**

**Egypt**
Shawky et al. (2005) described a 15-year-10-month-old female representing characteristic features of Martsolf syndrome. Her parents were first cousins and her young brother was normal. She had bad scholastic achievement. At the age of six years, she represented bilateral cataract extraction. On examination, she was short in stature, and she had subnormal mentality, receding forehead, hypertelorism, retromicrognathia, and clinodactyly. Her bone age corresponded to ten years. Ultrasound revealed small ovaries and uterus. FSH and LH levels were elevated, whereas estradiol level was below normal.

**References**

**Contributors**
Abeer Fareed: 17.7.2006