



## McDonough Syndrome

### WHO International Classification of Diseases

Congenital malformations, deformations and chromosomal abnormalities

### OMIM Number

248950

### Mode of Inheritance

Autosomal Recessive

### Description

McDonough syndrome is an autosomal recessive inherited disorder originally described by Neuhaüser and Optiz in 1975. The main features of this of this syndrome include psychomotor retardation, congenital heart disease, a malformed thorax and peculiar facies consisting of prominent supraorbital ridges synorphy, strabismus, a large nose, malocclusion of teeth and large inverted ears.

### Epidemiology in the Arab World

#### United Arab Emirates

In 1996, Al-Gazali et al. reported four sibs from a consanguineous Bedouin family of Arabic descent from the United Arab Emirates, sharing some features in common with McDonough syndrome, and in addition had a short webbed neck, skin and nail changes. The four affected sisters were between 4 to 6 years old and showed a striking facial appearance associated

with short stature, mental retardation, macrocephaly, a short webbed neck, incurved little fingers and chest deformity. In addition they had dry skin with florid eczema in one of them and dystrophic nails in another. In two of them there was an associated congenital heart defect. The facies were long with long forehead, arched eyebrows, hypoplastic supraorbital ridges, a downward eye slant, a large nose, and low set ears with thick helix. The family also had six unaffected children, a history of one miscarriage and one male child who had a large head and similar features of the four sibs in this report, who died at the age of 5 months of unknown cause. The father had a sister who was said to be short and mentally retarded with similar facial features. Al-Gazali et al. (1996) proposed that the constellation of abnormalities in the affected children could represent a newly recognized syndrome that overlaps with McDonough syndrome.

### References

Al-Gazali LI, Aziz SA, Salem F. A syndrome of short stature, mental retardation, facial dysmorphism, short webbed neck, skin changes and congenital heart disease. *Clin Dysmorphol.* 1996; 5(4):321-7.

### Contributors

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