



Anophthalmos with Limb Anomalies

Alternative Names

Waardenburg Anophthalmia Syndrome
Anophthalmos-Syndactyly
Ophthalmocromelic Syndrome
Waardenburg Recessive Anophthalmia Syndrome
Anophthalmia-Waardenburg Syndrome
Anophthalmos-Limb Anomalies Syndrome
Crooked Fingers Syndrome
Syndactyly-Anophthalmos Syndrome

WHO International Classification of Diseases

Congenital malformations, deformations and chromosomal abnormalities

OMIM Number

206920

Mode of Inheritance

Autosomal recessive

Description

Waardenburg Anophthalmia Syndrome (WAS) was reported in 18 families showing a male:female ratio of 1:3. Consanguinity was noted in 90% of the families. The most common clinical abnormalities associated with WAS were anophthalmia (absence of the eye) and foot malformations, which were occurring in 90% of patients. Some shortness of the palpebral fissures and smallness of the bony orbits were found to be associated with ophthalmia. The most common and distinctive foot abnormality is the presence of only four toes bilaterally. Most studied cases showed a wide gap between the 1st and 2nd toes. Joint laxity, club foot, valgus deformity, hypoplastic fibula, bowed tibia, and hip dislocation were observed in some cases. About 75% of the cases had abnormalities in both hands with the most distinctive abnormality of basal synostosis of the fourth and fifth metacarpals. Also, camptodactyly affecting the second through fifth fingers was noted in several cases. About 50% of the patients showed postnatal growth

delay. Severe mental retardation was also present in 50% of the reported cases.

Molecular Genetics

Waardenburg Anophthalmia Syndrome (WAS) is a distinct syndrome and is transmitted as an autosomal recessive trait. Also, the syndrome is thought to be familial.

Epidemiology in the Arab World

Lebanon

Megarbane et al. (1998) studied a 12-day-old boy who presented ophthalamo-acromelic syndrome (OAS). He was the first child of healthy consanguineous Lebanese parents. Ultrasound was performed at 21 weeks of gestation showing an oligoamnios. The child displayed bilateral anophthalmos (no eyeballs), adherent eyelids, deeply set small orbits, anteverted nares, micrognathia, very short neck, and narrow chest. X-ray of the upper and lower limbs showed a syndactyly between the second and the third fingers with metacarpal polydactyly on the right hand, an oligodactyly with the presence of only three fingers on the left hand, and a rudimentary toe on the right foot. The subarachnoid spaces were moderately enlarged. The chromosomes were normal (46,XY). Megarbane et al. (1998) excluded other syndromes associated with anophthalmia/microphthalmia and limb defects due to the absence of skin lesion and visceral malformations, and the presence of unique limb defects and severe malformations of the eyes in their studied case. The syndactyly between the second and third fingers, a metacarpal polydactyly, and a lobster-claw hand deformity were firstly reported by Megarbane et al. (1998) which might expand the phenotype of OAS. The variable phenotypes of the limb and eye malformations were thought to be caused by mutations at single locus.



References

Megarbane A, Souraty N, Tamraz J.
Ophthalmo-acromelic syndrome
(Waardenburg) with split hand and
polydactyly. Genet Couns. 1998; 9(3):195-9.

Contributors

Abeer Fareed: 21.6.2006

