



Acrocephalopolysyndactyly Type II

Alternative Names

ACPS II

Carpenter Syndrome

WHO International Classification of Diseases

Congenital malformations, deformations and chromosomal abnormalities

OMIM Number

201000

Mode of Inheritance

Autosomal recessive

Description

Acrocephalopolysyndactyly is a group of several syndromes, (Apert syndrome (Acrocephalosyndactyly, Type I), Carpenter syndrome (Acrocephalosyndactyly, Type II), Saethre-Chotzen syndrome (Acrocephalosyndactyly, Type III), and Pfeiffer syndrome (Acrocephalosyndactyly, Type V) whose basic features are craniosynostosis and polysyndactyly. Carpenter syndrome is of autosomal recessive inheritance and classically consists of acrocephaly with variable synostosis of sagittal, lambdoid and coronal sutures. The most common clinical features of the syndrome are syndactyly, polydactyly, especially preaxial polydactyly of toes, congenital heart disease, mental retardation, hypogonadism, obesity, umbilical hernia, coxa vara, pes varus and clinodactyly. The clinical spectrum and additional malformations in Carpenter syndrome may show considerable differences. Besides common clinical presentation, different skeletal abnormalities as genu valgum, lateral displacement of patella, poor development of acetabula, absent coccyx, spina bifida occulta and kyphoscoliosis may be seen. Dental developmental abnormalities, hypodontia, low-set ears or malformed ears, epicanthal folds, depressed nasal bridge and thick neck may also accompany the major clinical findings.

Epidemiology in the Arab World

Egypt

Temtamy (1966) described a male patient, born to first cousin parents, with Carpenter syndrome. He had preaxial polydactyly of the toes and closure of the coronal suture. Temtamy (1966) suggested an autosomal recessive inheritance for the syndrome. In fact, It is only after the description of Temtamy (1966) that Carpenter syndrome was recognized as a distinct disease entity.

Lebanon

[See also: Palestine > Der Kaloustian et al., 1972]

Palestine

In 1972, Der Kaloustian et al. described a Moslem Palestinian patient with acrocephalopolysyndactyly Type II.

In 1997, Zlotogora (1997) analyzed 2000 Palestinian Arabic families and found that in 98 families at least one individual had congenital hydrocephalus and/or open neural tube defect. In 22 families the brain malformation was part of a syndrome: Meckel syndrome in 10, Warburg syndrome in another 5, Carpenter in one, and undiagnosed in 6 families.

References

- Der Kaloustian VM, Sinno AA, Nassar SI. Acrocephalopolysyndactyly, type II (Carpenter syndrome). *Am J Dis Child.* 1972; 124(5):716-8.
- Temtamy SA. Carpenter's syndrome: acrocephalopolysyndactyly. An autosomal recessive syndrome. *J Pediatr.* 1966; 69(1):111-20.
- Zlotogora J. Genetic disorders among Palestinian Arabs. 2. Hydrocephalus and neural tube defects. *Am J Med Genet.* 1997; 71(1):33-5.



Contributors

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