Answers to Medical Quiz

Sheikha S Al Arrayed, MBChB, PhD*

A1. Carpenter syndrome: Acrocepalopolysyndactyly

A2. Apert Syndrome. Bardet-Biedl Syndrome

A3. Autosomal recessive inheritance

A4. Depends on the degree of mental retardation and severity of the congenital heart disease. Orthopaedic and cardiac correction should be considered when indicated.

Clinical Features: Acrocephaly, soft tissue syndactyly of the third and fourth fingers associated with brachymesophalangy, preaxial polydactyly and syndactyly of toes, congenital heart disease, hypogenitalism, mild obesity, and frequently mental retardation characterise the disorder.

Specific Diagnosis: Asymmetric premature synostosis of all cranial sutures produced a distorted calvaria. The nasal bridge is often flat and there may be dystopia canthorum. The hands are short with stubby fingers with syndactyly most marked between the third and fourth fingers. There is usually a single flexion crease. Congenital heart disease has been reported in several cases. Omphalocele, undescended testes and variable mental retardation complete the picture. Radiographically the proximal phalanx of the thumb has two ossification centers. Usually there is bilateral varus deformity of the feet and preaxial polydactyly with duplication of the first and second toe. The toes may exhibit soft tissue syndactyly, meta varus and reduplication of the second toe. In nearly all cases there has been genu valgum with lateral displacement of the patella.

Prenatal Diagnosis: By ultrasonography.

Pathogenesis: Unknown

Risk of recurrence for patient sibs: Mendelian inheritance, AR 25%.

Age of detectability: At birth.

Prevention: Unknown, genetic counseling indicated.

Detection of carrier: Unknown.

REFERENCES

1. Carpenter syndrome. Smiths recognizable patterns of Human malformation. 4th edn. 1988:370-1.
2. Temtamy SA. Carpenters syndrome Acrocephalopolysyndactyly, an autosomal recessive syndrome. Pediatric 1966;69:111.
3. Robinson LK, et al. Carpenter syndrome. Natural history and clinical spectrum. Am J Med Genet 1985;20:461.

* Consultant Clinical Geneticist
Paediatric Department
Salmaniya Medical Complex
State of Bahrain