Geçen Sayının Bilmece Olgusunun Yanıtı

CHARGE

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The diagnose of our patient was CHARGE association. The association has 7 components.[1,2]

* **C**olobomatous malformation sequence (80%)
* **H**eart anomaly (70%, septal defects and conotruncal abnormalities are the most frequent anomalies, others are patent ductus arteriosus and TOF)
* **A**tresia choanae (50%)
* **R**etarded growth and development and/or central nervous system anomalies (mental 70% and growth 75%)
* **G**enital anomalies and/or hypogonadism (male 70%, female 30%)
* **E**ar anomalies and/or deafness (90%)

Each of the main features of the syndrome can be present in varying degrees of severity ranging from near normal to the most severe, and no single feature is present in all individuals affected with the defects.[1,2] Other anomalies, including facial asymmetry, unilateral facial nerve paralysis, renal abnormality, orofacial clefts, micrognathia, Di George Sequence, ocular hypertelorism, anal atresia and/or stenosis, omphalocele, esophageal atresia, tracheoesophageal fistula, microcephaly, central nervous system anomalies, growth hormone deficiency, rib anomalies, ptosis, scoliosis and clinodactyly frequently accompany the main features.[1,2] To make a diagnosis, presence of at least 4 of the 7 features of the association is required, including a major anomaly (ie, choanal atresia, coloboma).[2-4] Our patient had ***bilateral choanal atresia, heart anomaly (TOF), intrauterine and postnatal growth retardation, antenatal polyhydramnios, velopharyngeal discoordination, micrognathia, high-arched palate, dysmorphic facies and ear anomaly.***

Antenatal polyhydramnios, intrauterine growth retardation, antenatal heart and brain abnormality, respiratory distress and/or apnea, central cyanosis, dysmorphic features, swallowing and feeding difficulty during neonatal period should alert the physician for the syndrome. Criteria for poor survival include the following.[1-5]

- Bilateral choanal atresia
- Complex cyanotic congenital heart disease
- Central nervous system anomalies
- Esophageal atresia

Most incidents of CHARGE Syndrome are sporadic. A spesific gene defect has not been identified. A few individuals with inverted duplication of 14q22-q24.3, microdeletion of chromosome 22q.11.2, and 9p- chromosomal defect are reported to have CHARGE-like features.[5]

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