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## Case Report

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# A THREE-DAY-OLD FEMALE WITH CARPENTER'S SYNDROME

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**Carpenter's syndrome (acrocephalopolysyndactyly type II Carpenter)** is a rare syndrome characterized by acrocephaly, facial dysmorphism, and brachyclinosyndactyly of the hands and polysyndactyly of the feet.

To our knowledge, there are about 40 reported cases of Carpenter's syndrome. The existence of this syndrome in the siblings of consanguineous parents suggests an autosomal recessive inheritance. The case reported here was a three-day-old girl with many abnormalities diagnosed both clinically and at autopsy. It is noteworthy that Carpenter's syndrome had existed in more than one sibling in this family. Therefore, prenatal diagnosis of this syndrome by sonography during pregnancy is recommended.

*Archives of Iranian Medicine, Volume 7, Number 3, 2004: 222 – 224.*

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**Keywords:** Acrocephalopolysyndactyly • Carpenter's syndrome • congenital anomaly

### Introduction

Carpenter first described a syndrome in two sisters who had peculiar facies, synbrachydactyly and polysyndactyly of feet. Temtamy reviewed the findings of 12 similar cases in literature and added one of her own. She stated that the cardinal features of Carpenter's syndrome were different from those of Laurence-Moon-Biedl and Apert's syndromes.<sup>1</sup> Carpenter's syndrome is a very rare craniofacial condition. The cardinal features of this syndrome are acrocephaly, peculiar facies, and brachysyndactyly on fingers, preaxial polysyndactyly on feet, hypogenitalism, obesity, and mental retardation.<sup>1-5</sup> Thirty-three percent of the cases with Carpenter's syndrome are associated with congenital heart defects such as ventricular septal defect (VSD), atrial septal defect (ASD), patent ductus arteriosus (PDA), pulmonary artery stenosis, and tetralogy of Fallot.<sup>3</sup> Balci et al presented a case with transposition of great

vessels.<sup>1</sup> Superior vena cava duplication has also been reported.<sup>5,6</sup> Other manifestations which might rarely be observed include genu valgum, coxa vara, pes varus, and umbilical hernia.<sup>2,4</sup> To date, there are approximately 40 reported cases of Carpenter's syndrome in the literature.<sup>1</sup> We report the third child of a family with Carpenter's syndrome who had several external and internal anomalies including craniosynostosis, acrocephaly, syndactyly, polysyndactyly, clubfoot, VSD, and bicornuate uterus.

### Case Report

The case we are reporting was a three-day-old girl born in April 2001 by cesarean section at her 36<sup>th</sup> week of gestational age of a 27-year-old mother in Ghaem Hospital of Mashhad University, Mashhad, Iran. She was the third live conceptus of her mother after one stillbirth and one aborted fetus. Her mother and father were first cousins. The first gestation ended in a 40-week-old still birth fetus with features similar to our case. The second gestation was aborted at 18 weeks of gestational age. There were no histories of genetic or congenital anomalies in this family. Karyotypes

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**Figure 1.** Tower skull, frontal bossing, and epicanthal folds.

for her parents were found to be normal. The baby died after 3 days and was referred to the department of pathology for autopsy. On external examination, the body weight was 2,900 g and crown heel length was 44 cm. The head, chest, and abdominal circumferences were 32 cm, 31 cm, and 34 cm, respectively. Anterior and posterior fontanels were closed and the sutures were overriding. The patient was found to have a tower skull and a peculiar face characterized by frontal bossing, epicanthal folds, low set ears, posteriorly-rotated ears, and micrognathia (Figure 1). The neck was broad and short. The nipples were 3 mm in diameter (normal range, 5 to 10 mm) and an umbilical hernia was observed. The hands were short and membranous syndactyly was present. Moreover, polysyndactyly (six fingers) in each



**Figure 2.** Shows syndactyly in the hands and polysyndactyly in feet.



**Figure 3.** Gross specimen of the heart showing a ventricular septal defect of 1 cm in diameter.

foot and bilateral clubfoot deformity were observed (Figure 2). On internal examination, the right and left thoracic cavities contained approximately 5 mL of clear fluid and the pericardial sac contained 1 to 2 mL of brownish fluid. The 20-g heart had a normal external configuration but there was a ventricular septal defect in the muscular septum approximately 1 cm in diameter (Figure 3). Four accessory spleens around the hilum of the spleen and a bicornuate uterus were also found. There were no other anomalies in her internal organs. The 23-g right lung and 15-g left lung were reddish purple and sank in the water. On microscopic examination, some of the alveoli were atelectatic. In addition, hyaline membranes lined the respiratory bronchioles, alveolar ducts, and random alveoli. In some of the alveoli, there were keratinocytes. The cause of death in this case was respiratory distress syndrome due to aspiration of amniotic fluid.

## Discussion

Carpenter described a new syndrome in two sisters who had acrocephalopolysyndactyly and defined the associated congenital malformations of the syndrome in 1909. However, the entity was accepted as a distinct syndrome only after Temtamy's suggestion.<sup>1</sup> We presented a case who had craniosynostosis, acrocephaly, peculiar facies, short neck, hypoplastic nipples, umbilical hernia, membranous syndactyly in the hands, polysyndactyly in the feet, bilateral clubfoot and, VSD. These anomalies are similar to those

reported by the others.<sup>1-3</sup> Accessory spleens and bicornuate uterus was also observed in our case. These anomalies had not been reported previously in the literature.<sup>1, 2, 7, 8</sup> Since the inheritance mode of Carpenter's syndrome is autosomal recessive, genetic counseling is mandatory.<sup>8</sup> Since the case was the third child of the family, genetic counseling before pregnancy and prenatal examination by early ultrasonography at 12<sup>th</sup> to 14<sup>th</sup> weeks of gestation was recommended. Several authors reported Carpenter's syndrome in more than one sibling.<sup>3, 5</sup> However, there are also reports from sporadic cases. The interesting point with our case was the presence of more than one sibling with Carpenter's syndrome in her family.

### References

- 1 Balci S, Haytöglu T, Ozer S. A case of a four-day-old male with Carpenter's syndrome with transposition of great arteries. *Turk J Pediatr.* 1998; **40**: 461 – 466.
- 2 Balci S, Onol B, Eryilmaz M, Haytöglu T. A case of Carpenter's syndrome diagnosed in a 20-week-old fetus with postmortem examination. *Clin Genet.* 1997; **51**: 412 – 416.
- 3 Wiedemann HR, Kunze J, Grosse FR, Dibbern H. *Atlas of Clinical Syndromes: a Visual Aid to Diagnosis for Clinicians and Practicing Physicians.* 2nd ed. St. Louis: Mosby Year Book; 1992.
- 4 Gorlin RJ, Cohen MM, Levin LS. *Syndromes of the Head and Neck.* 3rd ed. New York: Oxford University Press; 1990.
- 5 Baraitser M, Winter RM. *Color Atlas of Congenital Malformation Syndromes?* London; Baltimore: Mosby-Wolfe; 1996.
- 6 Cohen DM, Green JG, Miller J, Gorlin RJ, Reed JA. Acrocephalopolysyndactyly type II—Carpenter's syndrome: clinical spectrum and an attempt at unification with Goodman and Summit syndromes. *Am J Med Genet.* 1987; **28**: 311 – 324.
- 7 Cohen MM Jr. Craniosynostosis and syndromes with raiosynostosis: incidence, genetics, penetrance, variability, and new syndrome updating. *Birth Defects Orig Artic Ser.* 1979; **15**: 13 – 63.
- 8 Piussan C, van Poperinghe M, Grumbach Y, Audebert M, Helsemans C, Risbourg B. Carpenter's syndrome [in French]. *Arch Fr Pediatr.* 1977; **34**: 891 – 898.