ORIGINAL ARTICLE

The 58th Case of Toriello-Carey Syndrome: The Association with Colpocephaly on Brain Magnetic Resonance Imaging and Computerized Tomography Scan

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ABSTRACT

Background: Toriello-Carey syndrome is a very rare clinical syndrome characterized by agenesis of the corpus callosum, congenital heart defect, telecanthus, short palpebral fissures, micrognathia or retrognathia, a small nose, and other congenital abnormalities. The syndrome was first described by Toriello and Carey from Michigan in 1988. Toriello and Carey reported the occurrence of the condition in four children; three of them were siblings and considered the syndrome to have an autosomal recessive inheritance. The aim of this paper is to describe the first case number 58 of this syndrome which is associated with colpocephaly and it is the first case in Iraq and in the Arab. **Materials and Methods:** An Iraqi female neonate with Toriello-Carey syndrome presenting with neonatal seizures is described. She had facial dysmorphism including telecanthus, narrow and short palpebral fissures, low-set ears, and retrognathia. She also had congenital heart defects. Radiologic brain abnormalities included complete agenesis of the corpus callosum with colpocephaly. **Conclusion:** The patient described in this report represents the 58th case of Toriello-Carey syndrome, and is the first case in Iraq and in the Arab. The case was associated with a unique radiologic, colpocephaly on brain magnetic resonance imaging and computerized tomography scan.

Key words: Agenesis of the corpus callosum, Arab, colpocephaly, Toriello-Carey syndrome

INTRODUCTION

Toriello-Carey syndrome is a very rare clinical syndrome characterized by agenesis of the corpus callosum, congenital heart defect, telecanthus, short palpebral fissures, micrognathia or retrognathia, a small nose, and other congenital abnormalities. The syndrome was first described by Toriello and Carey from Michigan in 1988. Toriello and Carey reported the occurrence of the condition in four children; three of them were siblings and considered the syndrome to have an autosomal recessive inheritance.[1]

Fifty-seven cases of the syndrome were reported, including the four patients reported by Toriello and Carey, one patient reported by Lacombe *et al*., one patient reported by Camera *et al*., one patient reported by Till *et al*., two patients reported by Chinen *et al*., one patient reported by Imaizumi, one patient reported by Aftimos and McGaughran, one patient reported by Wegner and Hersh, two patients reported by Paladini *et al*., two patients reported by Barisic *et al*., one patient reported by Kataoka *et al*., 25 patients reported by Toriello *et al*., one patient reported by Tegay *et al*., one patient reported by Uras *et al*., two patients reported by McGoey *et al*., one patient reported by Said *et al*., one patient reported by Maretti *et al*., one patient reported by El-Chammas *et al*., one patient reported by Yokoo *et al*., one patient reported by Tirali *et al*., one patient reported by China Xie *et al*., and two patients reported by Dikow *et al*.[1-25]

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The aim of this paper is to describe the first case number 58 of this syndrome, which is associated with colpocephaly and it is the first case in Iraq and in the Arab.

MATERIALS AND METHODS

A 20-day-old female neonate who was the second child to consanguineous parents who were hospitalized due to seizures in the department of pediatrics was referred after control of seizure with carbamazepine and phenobarbitone due to some facial dimorphism. The girl had telecanthus, narrow and short palpebral fissures, low-set ears, and retrognathia [Figure 1]. Brain ultrasound and abdominal ultrasound showed normal findings, but brain magnetic resonance imaging (MRI) and brain computerized tomography (CT) scan showed complete agenesis of the corpus callosum with colpocephaly. Echocardiography showed small subaortic ventricular septal defect, small muscular ventricular septal defect, secundum atrial septal defect, mild pulmonary stenosis, and mild coarctation of the aorta. Laboratory testes excluded metabolic abnormalities such as hypoglycemia and hypocalcemia as a cause of seizures.

DISCUSSION

Toriello-Carey syndrome is a very rare clinical syndrome characterized by agenesis of the corpus callosum, congenital heart defect, telecanthus, short palpebral fissures, micrognathia or retrognathia, a small nose, and other congenital abnormalities. The syndrome was first described by Toriello and Carey from Michigan in 1988. Toriello and Carey reported the occurrence of the condition in four children; three of them were siblings and considered the syndrome to have an autosomal recessive inheritance.[1]

Camera *et al*. from Italy reviewed the previous five cases and reported the sixth case of the syndrome.[3]

Figure 1: (a and b) The girl had telecanthus, narrow and short palpebral fissures, low-set ears, and retrognathia

Czarnecki *et al*. from the USA reported two patients with Toriello-Carey syndrome. The first patient was a male who had micrognathia, cleft soft palate, hypoplastic right ear, anotia on the left side, cerebellar vermis hypoplasia, hydrocephalus, agenesis of the corpus callosum, and hypoplastic left heart. He died 2 days after birth. The second patient was the male sibling of a patient reported previously (Am J Med Genet 42: 374–376; 1992). He had large fontanelles, telecanthus, a short nose, small and malformed ears, micrognathia, a large ventricular septal defect, and pulmonary stenosis. At age 8 months, he had growth retardation and developmental delay. A sister is unaffected.[4]

Czarnecki *et al*. reviewed eight other patients with Toriello-Carey syndrome, six of them were male. The two female patients were less severely affected and were still alive. Of the other male patients, all are deceased except one who is still alive at age 5 years; he has severe growth retardation, mental retardation, severe speech delay, and characteristic anomalies. Czarnecki *et al*. thought that the predominance of affected males and the milder phenotype in the female patients may suggest an X-linked gene or sex-influenced gene.[4]

Chinen *et al*. from Japan reported two Japanese sisters with a severe Toriello-Carey syndrome. The younger sister died suddenly at age 4 months. They emphasized that parental consanguinity in their cases supported and autosomal recessive inheritance of Toriello-Carey syndrome.[6]

Ohta *et al*. reported a patient with Toriello-Carey syndrome who had endocardial fibroelastosis.[7]

Aftimos and McGaughran from New Zealand reported a female patient with Toriello-Carey syndrome who had some additional findings including an anteriorly placed anus. The patient had a severe manifestation of complicated congenital heart disease, died in the neonatal period. Aftimos and McGaughran thought that the finding in their case was against the notion of the likelihood that the syndrome has an X-linked inheritance with more severe manifestations in males.[8]

Paladini *et al*. from Italy reported the first prenatal ultrasound diagnosis of the Toriello-Carey syndrome during the second trimester of pregnancy based on the detection of agenesis of the corpus callosum and spongious cardiomyopathy in a 22-week-old fetus of a couple with positive family history. The first child was diagnosed with Toriello-Carey syndrome at 1 year of age and had the typical facial anomalies, agenesis of the corpus callosum, and spongious cardiomyopathy.[11]

Kataoka *et al*. from Japan reported a boy with Toriello-Carey syndrome who had some additional findings, including a severe respiratory failure and intestinal dysmotility. The boy died of these two disorders at the age of 13 months.[13]

Toriello *et al*. from the USA reviewed 11 papers reporting 16 children in addition to the original four patients who reported by Toriello and Carey in 1988. Toriello *et al*. studied other 25 unpublished patients making the total number of the reported cases of the syndrome 45.[14]

Tegay *et al*. from the USA reported a patient with classical features of Toriello-Carey syndrome who an apparently balanced *de novo* translocation between chromosomes 2 and 14 [46, XY,t(2;14)(q33;q22)].[16]

Maretti *et al*. from Brazil reported a 13-year-old male with Toriello-Carey syndrome and leukoderma. Said *et al*. from Malta reported a girl with Toriello-Carey syndrome who had agenesis of the corpus callosum, a large cleft palate, telecanthus, hypertelorism, atrial septal defect, ventricular septal defect, and patent ductus arteriosus.[19]

El-Chammas *et al*. from the USA reported the second patient with Toriello-Carey syndrome and pancreatic insufficiency.[21]

Yokoo *et al*. from Japan reported a Japanese boy with Toriello-Carey syndrome who had severe congenital tracheal stenosis who was managed with respiratory care without surgical tracheal plasty and the patient to be alive at 18 months of age.[22]

The syndrome was reported from many countries in the world including the USA (at least ten patients), Japan (five patients), Italy (two patients), Croatia (two patients), Germany (two patients), Turkey (two patients), France, New Zealand, Brazil, Malta, and China.[1-25]

However, the syndrome has not been reported in the Arab or Africans.

CONCLUSION

The patient described in this report represents the 58th case of Toriello-Carey syndrome, and is the first case in Iraq and in the Arab. The case was associated with a unique radiologic, colpocephaly on brain MRI and CT scan.

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REFERENCES

1. Toriello HV, Carey JC. Corpus callosum agenesis, facial anomalies, Robin sequence, and other anomalies: A new autosomal recessive syndrome? Am J Med Genet 1988;31:17-23.
2. Lacombe D, Creusot G, Battin J. New case of Toriello-Carey syndrome. Am J Med Genet 1992;42:374-6.
3. Camera G, Righi E, Romagnoli G. Toriello-Carey syndrome: Report of a new case. Clin Dysmorphol 1993;2:260-3.
4. Czarnecki P, Lacombe D, Weiss L. Toriello-Carey syndrome: Evidence for X-linked inheritance. Am J Med Genet 1996;65:291-4.
5. Till M, Bourgeois J, Plauchu H. Toriello-Carey syndrome. Am J Med Genet 1997;70:332.
6. Chinen Y, Tohma T, Izumikawa Y, Taketomi H, Iha T, Ohta T, *et al*. Two sisters with Toriello-Carey syndrome. Am J Med Genet 1999;87:262-4.
7. Ohta H, Masuno M, Kimura J, Imaizumi K, Kuroki Y, Yasui S, *et al*. Toriello-Carey syndrome with endocardial fibroelastosis. Am J Med Genet 1999;87:271-2.
8. Aftimos S, McGaughran J. Toriello-Carey syndrome: Case report with additional findings. Am J Med Genet 2001;98:273-6.
9. Wegner KJ, Hersh JA. Toriello-Carey syndrome: An additional case and summary of previously reported cases. Clin Dysmorphol 2001;10:145-8.
10. Imaizumi K. [Toriello-Carey syndrome]. Ryoikibetsu Shokogun Shirizu 2001;34:737-8.
11. Paladini D, Russo MG, Tartaglione A, Loffredo A, Martinelli P. Prenatal ultrasound diagnosis of Toriello-Carey syndrome. Prenat Diagn 2002;22:1185-7.
12. Barisic I, Peter B, Mikecin L. Further delineation of the Toriello-Carey syndrome: A report of two siblings. Am J Med Genet A 2003;116A:188-91.
13. Kataoka TR, Ito A, Nakabori T, Koma Y, Nakai H, Kamata S, *et al*. Toriello-Carey syndrome associated with respiratory failure and non-mechanical ileus. Am J Med Genet A 2003;120A:537-41.
14. Toriello HV, Carey JC, Addor MC, Allen W, Burke L, Chun N, *et al*. Toriello-Carey syndrome: Delineation and review. Am J Med Genet A 2003;123A:84-90.
15. Martin-Denavit T, Till M, Plauchu H. Toriello-Carey syndrome and unbalanced translocation t(8;18)(p12;q22). Am J Med Genet A 2004;128A:219-21.
16. Tegay DH, Chan KK, Leung L, Wang C, Burkett S, Stone G, *et al*. Toriello-Carey syndrome in a patient with a de novo balanced translocation [46,XY,t(2;14)(q33;q22)] interrupting SATB2. Clin Genet 2009;75:259-64.
17. Uras N, Sandal G, Oguz S, Aydemir O, Erdeve O, Dilmen U. Toriello-Carey syndrome in a Turkish newborn. Genet Couns 2009;20:243-7.
18. McGoey R, Varma A, Lacassie Y. Siblings with phenotypic overlap with Toriello-Carey syndrome and complex cytogenetic imbalances including 3q29 microduplication and 6p25 microdeletion: Review of the literature and additional evidence for genetic heterogeneity. Am J Med Genet A 2010;152A:3068-73.
19. Maretti MB, Haddad AS, Ferreira MC, Guaré Rde O, Alonso LG. Toriello Carey syndrome: Genetic, clinical, and oral considerations: A case report. Spec Care Dentist 2011;31:68-72.
20. Said E, Cuschieri A, Vermeesch J, Fryns JP. Toriello-Carey syndrome with a 6Mb interstitial deletion at 22q12 detected by array CGH. Am J Med Genet A 2011;155A:1390-2.
21. El-Chammas KI, Venkatasubramani N, Veith R, Sekhri N, Rhead W, Toriello HV, *et al*. Pancreatic insufficiency in

Toriello-Carey syndrome: Report of a second patient. Am J Med Genet A 2012;158A:1208-11.
22. Yokoo N, Marumo C, Nishida Y, Iio J, Maeda S, Nonaka M, *et al*. A case of Toriello-Carey syndrome with severe congenital tracheal stenosis. Am J Med Genet A 2013;161A:2291-3.
23. Tirali RE, İlhan B, Şar Ç, Çehreli SB. Dental and dentofacial problems in a female child with Toriello-Carey syndrome: Changes in 3 years. Spec Care Dentist 2016;36:288-90.
24. Xie L, Luo X, Yang J, Wang J, Nie C, Wang Z. A Chinese patient with Toriello-Carey syndrome and an interstitial deletion of 3q. Am J Med Genet A 2017;173:721-6.
25. Dikow N, Granzow M, Graul-Neumann LM, Karch S, Hinderhofer K, Paramasivam N, *et al*. DDX3X mutations in two girls with a phenotype overlapping Toriello-Carey syndrome. Am J Med Genet A 2017;173:1369-73.

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