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Taiwanese Journal of Obstetrics & Gynecology 50 (2011) 528–530

www.tjog-online.com

Research Letter

Prenatal diagnosis of a *de novo* interstitial deletion of chromosome 20q12 in a fetus with complex congenital heart defects, corpus callosum agenesis and intrauterine growth restriction

Yi-Hui Lin [a], Chih-Ping Chen [a,b,c,d,e,*], Tze-Chien Chen [b], Chun-Sen Hsu [a], Szu-Yuan Chou [a], Shu-Chin Chien [f]

[a] *Department of Obstetrics and Gynecology, Taipei Medical University-Wan Fang Hospital, Taipei, Taiwan*
[b] *Department of Obstetrics and Gynecology, Mackay Memorial Hospital, Taipei, Taiwan*
[c] *Department of Medical Research, Mackay Memorial Hospital, Taipei, Taiwan*
[d] *Department of Biotechnology, Asia University, Taichung, Taiwan*
[e] *School of Chinese Medicine, College of Chinese Medicine, China Medical University, Taichung, Taiwan*
[f] *Departments of Medical Genetics, and Obstetrics and Gynecology, China Medical University Hospital, Taichung, Taiwan*

Accepted 12 May 2010

A 22-year-old, gravida 2, para 1 patient was referred to the hospital at 26 gestational weeks, because of fetal anomaly. The parents were non-consanguineous and healthy. The mother denied any exposure to alcohol, teratogenic agents, irradiation or infectious diseases during this pregnancy. Level II ultrasonography at 26 gestational weeks, revealed a growth-restricted fetus with a biparietal diameter of 5.9 cm (equivalent to 23 gestational weeks), and abdominal circumference of 17.4 cm (equivalent to 23 gestational weeks), a femur length of 3.9 cm (equivalent to 23 gestational weeks), atrioventricular septal defects and corpus callosum agenesis. Genetic amniocentesis revealed an interstitial deletion of the band 20q12, or 46,XY,del(20)(q11.2q13.1) (Fig. 1). The parental karyotypes were normal. Despite genetic counseling of an unfavorable outcome, the parents opted to continue the pregnancy. The proband was delivered at 38 weeks' gestation with a birth body weight of 2050 g (< 5th centile), body length of 44 cm (< 5th centile) and head circumference of 29 cm (< 5th centile). The Apgar scores were 5 and 7 at 1 and 5 minutes, respectively. At birth, the male baby manifested low set ears, hypertelorism, epicanthal folds, a broad and flat nasal bridge, anteverted nostrils, and cleft palate. Postnatal echocardiography showed atrioventricular septal defects, tricuspid regurgitation, and patent ductus arteriosus. He underwent surgical corrections for the complex heart defects but died of postoperative complications at the age of 7 months.

Constitutional deletions of the long arm of chromosome 20 are very rare. This is the first report of prenatal diagnosis of

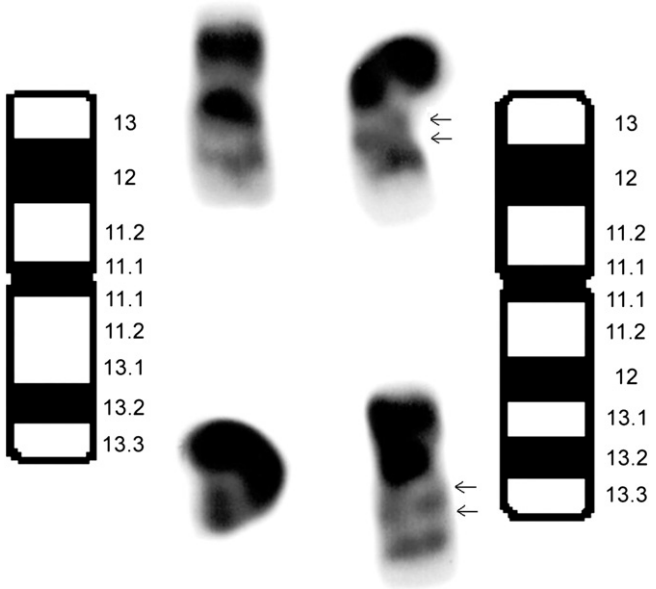

Fig. 1. Partial G-banded karyotype and ideogram of chromosome 20 and del(20)(q11.2q13.1).

* Corresponding author. Department of Obstetrics and Gynecology, Mackay Memorial Hospital 92, Section II, Chung-Shan North Road, Taipei, Taiwan.
E-mail address: cpc_mmh@yahoo.com (C.-P. Chen).

doi:10.1016/j.tjog.2011.10.024

Table 1
The clinical features of patients with chromosome 20q deletions.

	Fraisse et al. [3] q13.11-qter	Petersen et al. [5] q11.23-q13.11	Shabtai et al.[6] q13.11-q13.33 (mosaic)	Aldred et al. [2]		Geneviève et al.[4]		The present case q11.2-q13.1
				Patient 1 q13.31-q13.32	Patient 2 q13.13-q13.33	Patient 1 q13.2-q13.3	Patient 2 q13.2-q13.3	
Psychomotor retardation	+	+	+	+	+	+	+	−
Prenatal growth restriction	−	−	−	+	−	+	+	+
Postnatal growth restriction	+	+	−	+	+	+	+	+
Microcephaly	+	−	−	+	−	+	+	−
Short neck	+	−	+	+	−	−	−	−
Flat occiput	+	−	+	−	−	−	−	−
Hypertelorism	+	−	−	−	−	+	+	+
Epicanthal folds	+	−	−	−	−	−	−	+
Pterygium of eyes	+	−	+	−	−	−	−	−
Strabismus	−	+	−	−	−	−	−	−
Anteverted nostrils	+	−	−	−	−	−	−	+
Bulbous nose	+	−	−	+	−	−	+	−
Broad nasal bridge	+	−	+	+	−	−	+	+
High arched palate	+	−	−	−	−	−	−	−
High forehead	+	−	−	−	−	−	+	−
Hirsutism	+	−	−	−	−	−	−	−
Thin upper lip	+	−	−	−	−	−	+	−
Micrognathia	+	−	−	−	−	−	+	−
Prognathism	−	−	+	−	−	−	−	−
Low set and malformed ears	−	+	−	−	−	−	+	+
Long philtrum	+	−	−	−	−	−	−	−
Macrostomia	−	−	−	−	−	−	−	−
Malformed hands/feet	+	−	+	+	+	−	−	−
Heart murmur	−	+	−	−	−	−	−	−
Cardiac malformation	−	+	−	−	−	−	−	+
Seizures	+	−	−	−	−	−	−	−
ADA deficiency	+	−	−	−	−	−	−	−
CNS malformation	−	−	−	−	−	−	−	+
Intractable feeding difficulties	−	−	−	+	−	+	+	−
Inguinal hernia	−	−	−	−	−	+	−	−
Undescended testes	−	−	−	−	−	+	−	−
Parental origin	Unknown	Unknown	Unknown	Paternal	Maternal	Paternal	Paternal	Unknown

ADA = adenosine deaminase; CNS = central nervous system.

a *de novo* interstitial deletion of chromosome 20q12. Acquired deletions of the long arm of chromosome 20 have been found in several hematological conditions, such as myeloproliferative disorders, myelodysplastic syndrome and acute myeloid leukemia [1]. Only seven cases of constitutional chromosome 20q deletions have been reported postnatally [2–6]. Four of seven cases had limb deformities. The clinical features of these patients are summarized in Table 1. Petersen et al. [5] first described an interstitial deletion of chromosome 20q12 in a 3-year-old boy with growth and developmental retardation, low set ears, broad nasal bridge, macrostomia, strabismus, and heart murmur. The mutation of *SLC2A10* located at 20q13.1, has been postulated to cause arterial tortuosity syndrome, which comprises generalized tortuousity and elongation of all major arteries, soft skin, joint laxity, severe keratoconus, and diffuse tortuosity of the carotids and of intracranial arteries [7]. Deficiency of the facilitative glucose transporter *GLUT10* encoded by *SLC2A10*, is associated with upregulation of the *TGF-beta* pathway in the arterial wall [8]. Our case is associated with atrioventricular septal defects, tricuspid regurgitation, patent ductus arteriosus, and corpus callosum agenesis, and provides evidence that genes at 20q12 may be responsible for the development of the heart and the brain.

Acknowledgments

The authors thank Professor Teresa L. Yang-Feng for her critical review, support, and encouragement.

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