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Report of One Case with Congenital Vascular Ring

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Abstract

Congenital vascular rings is an unusual congenital condition and severely affects the survival and life quality of the patients. A 23-year-old gravid woman was referred for counseling at 24 weeks of gestation because of abnormal ultrasound findings of fetal congenital vascular ring. Fetal echocardiography showed a complete vascular ring with a right aortic arch (RAA), left ductus arteriosus (LDA) and left intracardia echogenicfocus. Conventional cytogenetic analysis revealed an apparent balanced reciprocal translocation between the distal segment of the long arm of a chromosome 5 and the long arm of chromosome 2: 46, XY, t (2;5) (q3.5; q31.1). This abnormal karyotype was detected in gravid woman. However, the microarray analysis on amniocytes using HumanCytoSNP-12 array detected 2.57-Mb deletion at 22q11.21. Metaphase fluorescence *in situ* hybridization (FISH) analysis on cultured amniocytes confirmed an interstitial 22q11.2 deletion. The fetus was died owing to breathing and feeding difficulties. Our study highlights the clinical value of genetic detection and prenatal diagnosis of Congenital vascular rings by karyotype analysis coupled with SNP array.

Keywords: Congenital vascular ring; Prenatal genetic diagnosis; SNP array

Introduction

Congenital vascular rings is a group of congenital vascular anomalies with an estimated to be approximately 1%-2% in cardiovascular congenital anomalies [1]. It may present with severe symptoms of respiratory distress directly after birth or the development of milder symptoms and signs of tracheoesophageal compression later in life [2]. Vascular rings can occur in isolation or may be associated with other congenital heart defects as well as non-cardiac defects and chromosomal or genetic anomalies [3]. Currently, the diagnosis of vascular rings relies on prenatal ultrasound using the three-vessel trachea view [4-6]. But the standard prenatal genetic diagnosis is insufficient.

Here we report one case with congenital vascular ring. More importantly, our data could offer informative data for proper prenatal genetic counseling of pregnant women.

Case History

A 23-year-old gravid woman was referred for counseling at 24 weeks of gestation because of abnormal ultrasound findings of fetal congenital vascular ring. Fetal echocardiography showed a complete vascular ring with a right aortic arch (RAA), left ductus arteriosus (LDA) and left intracardia echogenicfocus. Other internal organs were unremarkable (Figure 1). Gravid woman had an abortion history. She was healthy and denied any recent infections or exposure to teratogens during this pregnancy. But her husband was diagnosed with congenital heart defects.

After obtaining informed consent, amniocentesis was performed for cytogenetic analysis and the microarray analysis. The fetal chromosomal result revealed an apparent balanced reciprocal translocation between the distal segment of the long arm of a chromosome 5 and the long arm of chromosome 2: 46,XY,t(2;5) (q3.5; q31.1) (Figure 2). Owing to the relatively poor data of this abnormal karyotype, this family chromosomal analysis was performed on the basis of G-banding technique at high resolution to explain this karyotype. This abnormal karyotype was detected in gravid woman and her father with normal phenotypes (Figure 3).

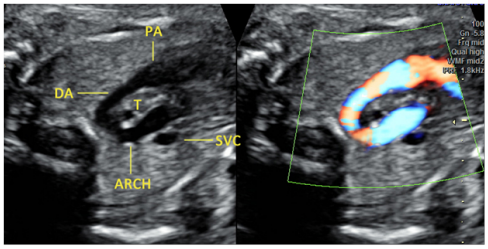

Figure 1: Echocardiograms obtained at 24 weeks' gestation in a fetus with vascular ring.

Figure 2: Fetal chromosome analysis by G-banding technique revealed 46, XY, t(2; 5) ($q^{3.5}$; q31.1) karyotype

The array analysis detected a 2.57 Mb deletion of the region 22q11.21 (chr22: 18,889,490-21,462,353) (Figure 4). According to the OMIM database the abnormal region presented here encompasses 32 disease-causing genes: *COMT, RTN4R, NOGOR, PRODH, PRODH2, SC2D4, GP1BB, BS, BDPLT1, SCARF2, SREC2, VDEG5, HCF2, HC2, SERPIND1, THPH10, L2TR1, SWNTS2, TBX1, DGS, CTHM, CAF5, TGA, DORV, VCF5, DGCR, SNAP29, CEDNIK, SLC25A1, SLC20A3, CTP* and *D2L2AD*. The parents requested repeated amniocentesis. *FISH*

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analysis on using Vysis DiGeorge region probe showed the presence of only one orange signal and two green signals, indicating a deletion of DiGeorge syndrome Tup-like enhancer of split 1 (TUPLE1) locus at 22q11.2 in the fetus (Figure 5).

We suggested terminating this pregnancy. But the couple selected to continue the pregnancy. After the fetus was born about 27 days, he was died owing to breathing and feeding difficulties (Figure 6).

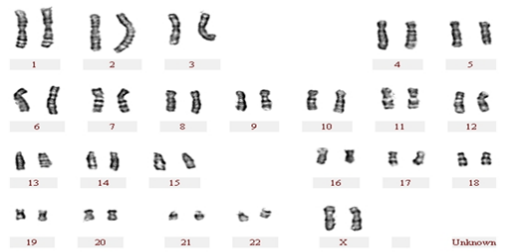

Figure 3: Gravid woman chromosome 46, XX, t(2; 5) ($q^{3.5}$; q31.1) karyotype.

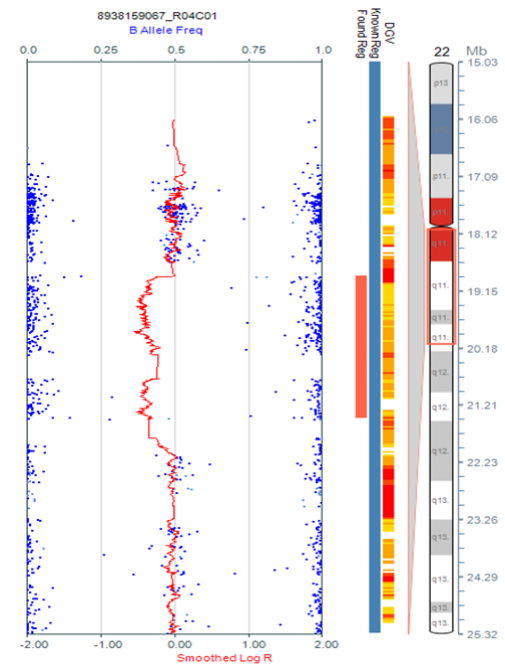

Figure 4: Chromosome microarray profile showing a 2.57 Mb loss of the region 22q11.21(18,889,490-21,462,353).

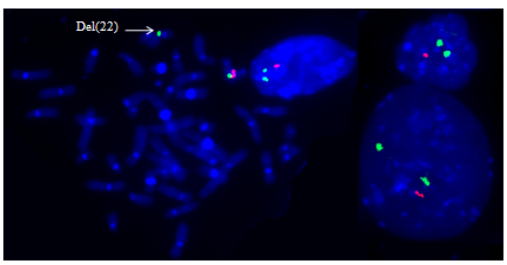

Figure 5: Fluorescence in situ hybridization analysis using Vysis LSI TUPLE 1 (HIRA)spectrum orange/LSI ARSA spectrum green probe set (Abbott Laboratories) shows a normal chromosome 22 (one orange signal and one green signal) and a del (22) chromosome (only one green signal) in a metaphase amniocyte.

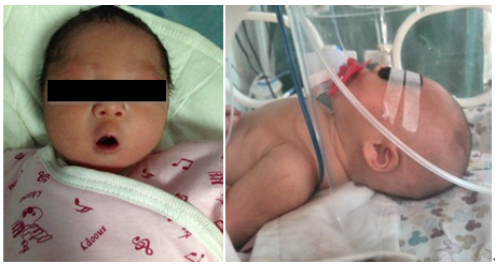

Figure 6: The outcome of this fetus at 27 days born.

Discussion

Certainly, vascular rings are diagnosed and managed operatively, outcomes are excellent. However, the literature regarding the prenatal diagnosis of vascular rings is extremely limited. Owing to vascular rings exhibit a wide spectrum of clinical severity, the severity of compression from the ring is unknown at the time of fetal diagnosis [7,8]. In some fetuses, vascular rings is identified which may progress at birth and evolve with ductal closure. It may be difficult to predict, making it hard to counsel parents about structural cardiac diseases. An optimal prenatal diagnosis program is desperately needed. Technologic advancements in fetal cardiac imaging have enhanced the capability for diagnosis of vascular rings in utero [9,10]. Several reports indicated the chromosomal abnormalities (particularlya 22q11.2 deletion) can be observed in patients with vascular rings, an association that is important for prenatal counseling [11-13]. In this cohort, we detected translocation t(2; 5) (q3.5; q31.1). The translocation between the short arm of chromosome 2 and the long arm of chromosome 5 have been reported and are associated with malignant histiocytosis [14]. Therefore, parental chromosomal analysis is important for appropriate genetic counseling in relation to an embryonic pregnancy. Our results showed that the mother's karyotype was 46,XX, t(2; 5) (q3.5; q31.1) and the father's karyotype was normal (data not shown). Based on the study observations, it seems that the fetal chromosomal karyotype was present by heredity. So this result demonstrated translocation t(2; 5) (q3.5; q31.1) was not a primary event in congenital vascular ring.

The karyotyping would have missed 66% of genomic abnormalities in their cohort. They propose to perform genomic high-resolution array testing assisted by pre-test counselling as a primary prenatal diagnostic test in cases of foetal ultrasound abnormalities. Prenatal genetic diagnosis after ultrasound detection of foetal abnormalities requires a fast diagnostic technique. prenatal SNP array testing is faster than karyotyping and allows detecting much smaller aberrations (~0.15 Mb) in addition to the microscopic unbalanced chromosome abnormalities detectable with karyotyping (~ >5 Mb) [15,16]. Although FISH and CGH array has also been successfully implemented into prenatal diagnosis, as we described before, we have chosen Illumina SNP array mainly because it requires only 50 ng DNA, long culturing can be avoided and rapid results can be provided within 72 hours. In this study, 2.57-Mb deletion at 22q11.21 was primarily diagnosed by SNP array. FISH detected a deletion of DiGeorge syndrome TUPLE1 locus at 22q11.2. Patients with 22q11.2 deletion syndrome can suffer from congenital heart diseases, palatal abnormalities, learning difficulties, immune deficiency, characteristic facial features, and hypocalcemia [17,18].

Conclusion

An increasing number of fetal vascular ring cases were detected by fetal echocardiography. It was crucial that those clinical doctors

explain it and offer a reasonable prenatal genetic counseling for family. We proposed several points. Firstly, congenital vascular rings should be diagnosed using the three-vessel trachea view and subsequent fetal echocardiography. Secondly, it was important step to know family history and choice fast diagnostic technique to assist prenatal genetic counseling. Finally, reasonable prenatal genetic counseling was offered for family including clinical severity, heredity and outcome.

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