Taiwanese Journal of Obstetrics & Gynecology 56 (2017) 697–699

Contents lists available at ScienceDirect

Taiwanese Journal of Obstetrics & Gynecology

journal homepage: www.tjog-online.com

Case Report

Dandy–Walker syndrome with duplex kidney abnormalities in trisomy 18 – A rare case report

Tun-Jun Wang [a], Yi-Ying Li [b], Wan-Ju Wu [b], Chi-Kang Lin [a], Chun-Kai Wang [a], Chen-Yu Wang [a], Kwei-Shuai Hwang [a], Her-Young Su [a, *]

[a] Department of Obstetrics and Gynecology, Tri-Service General Hospital, National Defense Medical Center, Taipei, Taiwan
[b] Taiji Fetal Medicine Center, Taipei, Taiwan

ARTICLE INFO

Article history:
Accepted 5 June 2017

Keywords:
Edwards syndrome
Dandy–Walker malformation
Trisomy 18

ABSTRACT

Objective: Trisomy 18 is one of the major numerical chromosomal disorders. The incidence of trisomy 18 is approximately one in 6000 live births. Dandy–Walker malformation (DWM) is the most common congenital malformation of the cerebellum, with an incidence of about one in 5000 live births. The incidence of trisomy 18 associated with DWM is rare and long-term survival rate is very low.
Case report: A case involving a 39-year-old pregnant female with a case of trisomy 18 associated with DWM.
Conclusion: The incidence of trisomy 18 associated with DWM is rare, and our report presents an unusual case that supplements our knowledge of this condition.
We report a case involving a 39-year-old pregnant female with a case of trisomy 18 associated with Dandy–Walker malformation (DWM). Fetal ultrasonography showed hypoplasia of the cerebellar vermis and dilatation of the fourth ventricle and was characterized by an enlarged posterior fossa. Fetal magnetic resonance imaging showed inferior vermian hypoplasia and a large posterior fossa cyst communicating with the fourth ventricle causing high insertion of the torcular herophili, which was compatible with DWM. Furthermore, the karyotyping report revealed trisomy 18. The incidence of trisomy 18 associated with DWM is rare, and our report presents an unusual case that supplements our knowledge of this condition.

Introduction

Trisomy 18 [1–3], also known as Edwards syndrome, is one of the major numerical chromosomal disorders. The incidence of trisomy 18 is approximately one in 6000 live births. Typical clinical manifestations of trisomy 18 include low birth weight, clenched hands with the index finger overlapping the third digit and the fifth digit overlapping the fourth, narrow hips with limited abduction, short sternum, rocker-bottom feet, microcephaly, prominent occiput, micrognathia, cardiac and renal malformations, and intellectual disability. About 92% of children with trisomy 18 die in the first year. About 25% of patients with full support may survive for one year. Survivors show significant neurodevelopmental delay [4].

Dandy–Walker malformation (DWM) was first described by Dandy and Blackfan in 1914 and is the most common congenital malformation of the cerebellum, with an incidence of about one in 5000 live births. The triad of DWM consists of (i) complete or partial agenesis of the vermis (ii) cystic dilatation of the fourth ventricle (iii) an enlarged posterior fossa with upward displacement of the lateral sinuses, tentorium, and torcular herophili. This triad can be found in association with supratentorial hydrocephalus. Systemic malformations associated with DWM may include cardiac anomalies (ventriculoseptal defects, patent ductus arteriosus, and transposition of the great arteries), urogenital anomalies (hydroceles, vesico-ureteral reflux, and abnormal kidney shape), and other abnormalities (duodenal atresia, cleft palate, and malformed limbs), which occur collectively in about half of the patients [5]. The etiology of DWM includes chromosomal abnormalities [6,7], single gene disorders, and exposure to teratogens [8].

The incidence of trisomy 18 associated with DWM is rare, and there are only five reports with 22 cases published from 1991 to

* Corresponding author. 5F, 325, Section 2, Cheng-Gong Road, Nei-Hu District, 114 Taipei, Taiwan.
E-mail address: su108868@gmail.com (H.-Y. Su).

http://dx.doi.org/10.1016/j.tjog.2017.08.022

2010 [9–13]. In Taiwan, there is no published report on trisomy 18 associated with DWM. Our report presents an unusual case that supplements our knowledge of this condition.

Case presentation

A 39-year-old pregnant female patient, gravida three para two, had no history of infection in the early pregnancy period. The patient did not undergo a chromosome test because she missed the scheduled period for amniocentesis. In addition, the fetus appeared small for gestational age, and polyhydramnios was observed since 28 weeks of gestational age. Fetal ultrasonography showed hypoplasia of the cerebellar vermis and dilatation of the fourth ventricle and was characterized by an enlarged posterior fossa (Fig. 1) at 33 weeks of gestational age. Fetal magnetic resonance imaging showed inferior vermian hypoplasia and a large posterior fossa cyst communicating with the fourth ventricle causing high insertion of the torcular herophili (Fig. 2), which was compatible with DWM. Furthermore, the image (Fig. 3) also showed left duplex kidney. Family history was unremarkable for congenital abnormalities, and both older siblings were healthy.

As the parents requested termination, the fetal heartbeat stopped after feticide with intracardiac KCl was injected at 36 weeks and two days of gestation. The female stillborn was delivered by vaginal delivery. She was 40 cm tall (shorter than −3.0 standard deviation, SD) and weighed 1680 g (lighter than −3.0 SD). Physical examination of the stillborn showed neither clenched hands nor rocker-bottom feet. The karyotyping report revealed trisomy 18. No autopsy was performed.

Discussion

There are many reports on the complications of chromosomal abnormalities with DWM; however, the incidence of trisomy 18 associated with DWM is rare. We found only 22 published cases of trisomy 18 complicated with DWM or Dandy–Walker variants. Imataka et al. summarized three reports regarding chromosome 18 and DWM [9]. These three reports show that trisomy 18 occurred in 20 of 78 cases with DWM. Lim et al. also presented a case report regarding ocular findings in a patient with trisomy 18 with a variant of Dandy–Walker syndrome [13].

In our case with trisomy 18 associated with Dandy–Walker syndrome, imaging examinations showed inferior vermian hypoplasia and a large posterior fossa cyst, which is compatible with DWM. Duplex kidney and aorta coarctation are both compatible with the common complicated deformation of DWM. Low birth weight is compatible with the features of trisomy 18; however, it is surprising that no other typical characteristics of trisomy 18, such as clenched hands or rocker-bottom feet, were noted.

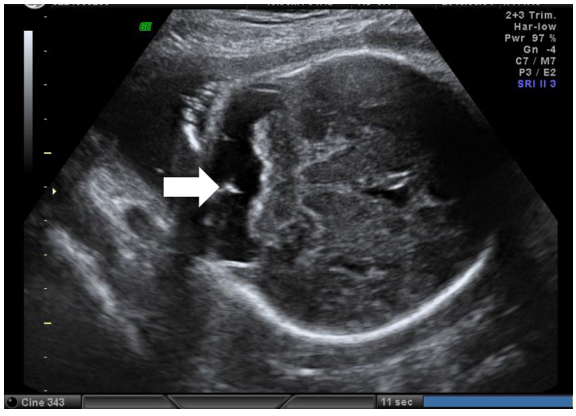

Fig. 1. Fetal ultrasonography showed enlarged posterior fossa (white arrow).

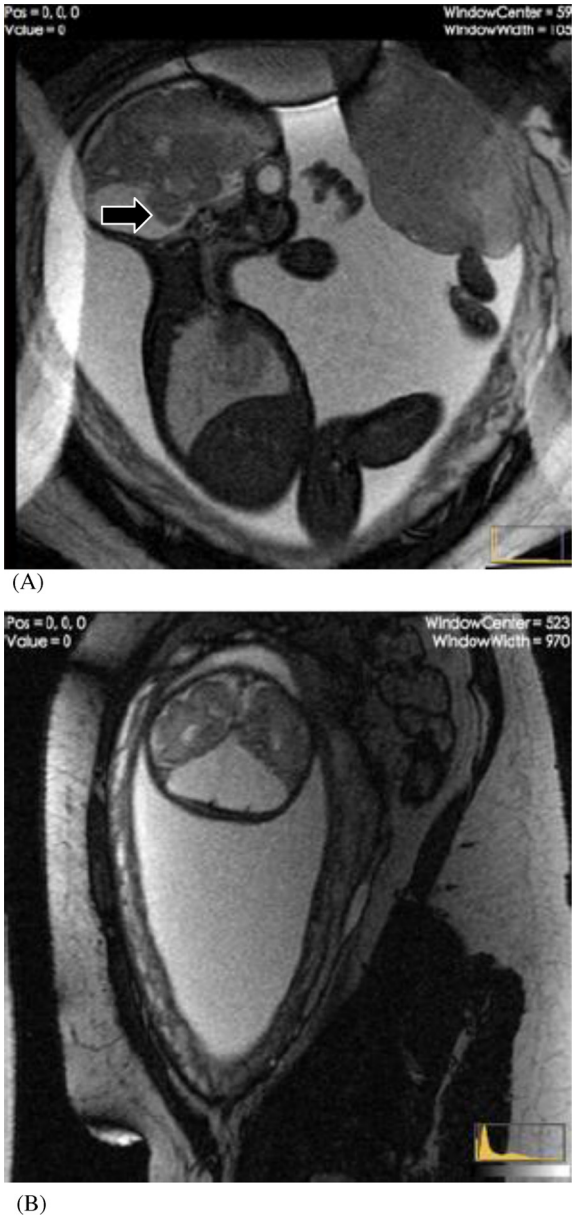

Fig. 2. (A) Fetal magnetic resonance imaging showed inferior vermian hypoplasia (black arrow) (B) and large posterior fossa cyst communicating with the fourth ventricle.

Because of the high risk of severe congenital cardiac disorders simultaneously occurring with trisomy 18, the long-term survival rate is very low. All of the published cases with trisomy 18 associated with DWM had a poor prognosis. In most cases, the pregnancy was terminated or the patient died at birth, and the maximal life span of the survivors was 414 days of age [9].

Due to the poor prognosis of DWM, it may occur as part of chromosomal abnormalities, single gene disorders, or exposure to teratogens. Therefore, routine karyotype investigation of the cases of DWM is warranted. If karyotypic abnormalities are identified, poor prognosis of the fetus may be estimated.

Ethics is an essential dimension for the management of pregnancies complicated with fetal anomalies. It is important to maintain an ethically justified balance between autonomy-based and beneficence-based obligations to the pregnant woman and

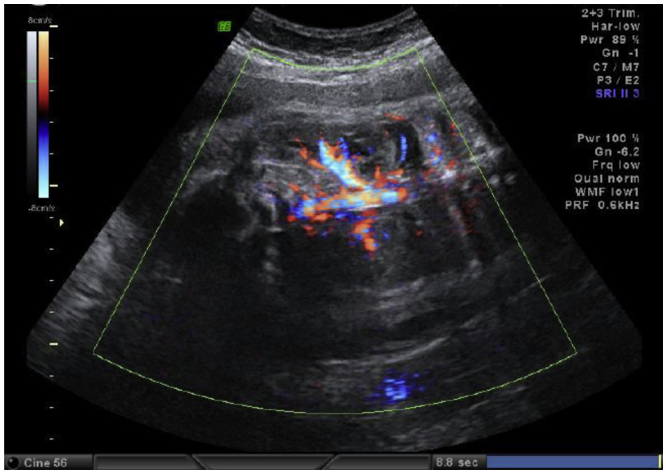

Fig. 3. Fetal ultrasonography showed duplex kidney, left.

beneficence-based obligations to the fetus [14]. Most professionals agree that feticide in cases of fetal anomaly will prevent parents and labor ward staff from facing the agony of neonatal distress and pain [15]. For the management of feticide in cases of fetal anomaly, respect for the pregnant woman's autonomy is the decisive concern.

In conclusion, the etiology of DWM includes chromosomal abnormalities, single gene disorders, and exposure to teratogens. Our case presented an unusual etiology of DWM that was associated with trisomy 18. The incidence of trisomy 18 associated with DWM is rare, and our report provides an unusual case that supplements our knowledge in the field of trisomy 18 and DWM.

Conflicts of interest

The authors declare that there is no conflict of interests regarding the publication of this paper.

References

[1] Chen CP, Su YN, Weng SL, Tsai FJ, Chen CY, Liu YP, et al. Rapid aneuploidy diagnosis of trisomy 18 by array comparative genomic hybridization using uncultured amniocytes in a pregnancy with fetal arachnoid cyst detected in late second trimester. Taiwan J Obstet Gynecol 2012;51:481–4.
[2] Chen CP, Wang LK, Chern SR, Kuo YL, Chen YN, Pan CW, et al. First-trimester diagnosis of recurrent omphalocele associated with fetal trisomy 18 but without parental mosaicism. Taiwan J Obstet Gynecol 2015;54:194–5.
[3] Chen CP, Hung FY, Chern SR, Wu PS, Chen YN, Chen SW, et al. Prenatal diagnosis of low-level mosaicism for trisomy 18 associated with a favorable fetal outcome. Taiwan J Obstet Gynecol 2016;55:902–3.
[4] Bacino CA, Lee B. Cytogenetics. In: Robert MK, editor. Nelson textbook of pediatrics. 20th ed. Philadelphia: Elsevier; 2016. p. 604–26.
[5] Millen KJ, Gleeson JG. Disorders of cerebellar and brainstem development. In: Swaiman K, Ashwal S, Ferriero D, Schor N, editors. Swaiman's pediatric neurology: principles and practice. 5th ed. Elsevier; 2012. p. 160–72.
[6] Tonni G, Lituania M, Chitayat D, Bonasoni MP, Keating S, Thompson M, et al. Complete trisomy 9 with unusual phenotypic associations: Dandy–Walker malformation, cleft lip and cleft palate, cardiovascular abnormalities. Taiwan J Obstet Gynecol 2014;53:592–7.
[7] Tan SJ, Chen CH, Chen CP, Chen CW, Chen CY, Hwang KS. Prenatal diagnosis of mosaic ring chromosome 15 with abnormal maternal serum Down syndrome screening and Dandy–Walker malformation. Taiwan J Obstet Gynecol 2012;51:109–11.
[8] Bacino CA, Lee B. Cytogenetics. In: Robert MK, editor. Nelson textbook of pediatrics. 20th ed. Philadelphia: Elsevier; 2016. p. 2802–18.
[9] Imataka G, Yamanouchi H, Arisaka O. Dandy–Walker syndrome and chromosomal abnormalities. Congenit Anom (Kyoto) 2007;47:113–8.
[10] Nyberg DA, Mahony BS, Hegge FN, Hickok D, Luthy DA, Kapur R. Enlarged cisterna magna and the Dandy–Walker malformation: factors associated with chromosome abnormalities. Obstet Gynecol 1991;77:436–42.
[11] Estroff JA, Scott MR, Benacerraf BR. Dandy–Walker variant: prenatal sonographic features and clinical outcome. Radiology 1992;185:755–8.
[12] Ulm B, Ulm MR, Deutinger J, Bernaschek G. Dandy–Walker malformation diagnosed before 21 weeks of gestation: associated malformations and chromosomal abnormalities. Ultrasound Obstet Gynecol 1997;10:167–70.
[13] Lim FF. Ocular findings in a case of trisomy 18 with variant of Dandy–Walker syndrome. Pediatr Neonatol 2010;51:292–5.
[14] Chervenak FA, McCullough LB, Skupski D, Chasen SD. Ethical issues in the management of pregnancies complicated by fetal anomalies. Obstet Gynecol Surv 2003;58:473–83.
[15] Peckham C. Feticide. In: Moody J, editor. Termination of pregnancy for fetal abnormality in England, Scotland and Wales. London: Royal College of Obstetricians and Gynaecologists; 2010. p. 29–31.