CASE REPORT

The crying sign: the winking umbilical cord

Aisling M Smith, David B Healy, C Anthony Ryan, Eugene M Dempsey

Department of Paediatrics and Child Health, University College Cork, Cork, Ireland

Correspondence to
Professor Eugene M Dempsey,
gene.dempsey@gmail.com

Accepted 3 March 2015

SUMMARY

A preterm baby girl, born at 34 weeks gestation, with features of Beckwith-Wiedemann syndrome was noted to have a relatively large umbilical stump. No fetal abnormalities had been detected on anatomy scan at 28 weeks and only mild polyhydramnios and macrosomia were noted on a 32-week ultrasound scan. Although there was no obvious omphalocele, clinical assessment of the umbilical cord revealed an abdominal wall defect through which bowel would protrude into the umbilicus when the infant was crying. In keeping with an abdominal wall defect α -fetoprotein was found to be elevated. Surgical consultation advised conservative management. Subsequently, detachment of the umbilical cord occurred 1 week postdischarge and a large umbilical hernia persists. Genetic analysis confirmed a diagnosis of Beckwith-Wiedemann syndrome.

Video 1 Protrusion of bowel into the umbilical cord through a small abdominal wall defect as the infant cries.

CASE PRESENTATION

A preterm baby girl born by spontaneous vaginal delivery at 34/40 gestation, weighing 2630 g was noted initially to have a relatively large umbilical cord stump. Antenatally, no fetal anomalies were detected on anatomy scan at 28 weeks and a 32-week ultrasound scan demonstrated mild polyhydramnios and macrosomia. Clinical assessment of the umbilical cord revealed the following (video 1). As she cried one could observe bowel protrusion into her umbilicus, the so-called winking umbilical cord. She also demonstrated features of macrosomia, macroglossia and mild hypotonia, suggesting a clinical diagnosis of Beckwith-Wiedemann syndrome.

OUTCOME AND FOLLOW-UP

Following surgical review of the abdominal defect, conservative management was advocated. Cranial, renal and abdominal ultrasound scans were normal and echocardiography revealed mild pulmonary branch stenosis. The umbilicus detached 1 week postdischarge and a large umbilical hernia persists. In keeping with abdominal wall defects α -fetoprotein was initially raised (16 502 μ g/L) but is falling with serial measurement. Genetic analysis confirmed a diagnosis of Beckwith-Wiedemann syndrome.

DISCUSSION

An omphalocele is a midline abdominal wall defect of variable size, with the herniated viscera covered by a membrane consisting of peritoneum on the inner surface, amnion on the outer surface and Wharton's jelly between the layers. The estimated

worldwide incidence of omphalocele ranges between 1.5 and 3 per 10 000 births, but varies regionally. The aetiology of omphalocele is unknown but is associated with advancing maternal age. Other potential risk factors include prematurity, alcohol consumption, tobacco, serotonin-reuptake inhibitors (SSRIs) and obesity in pregnancy. Omphaloceles are often associated with chromosomal abnormalities, in particular trisomies 13, 18 and 21, and with Beckwith-Wiedemann syndrome, which accounts for 10% of cases. There is a high incidence of associated anomalies (50–70%), including cardiac defects (30–50%).

Beckwith-Wiedemann syndrome affects 1 in 12 000 newborns and comprises features of macroglossia, organomegaly, early hypoglycaemia, an increased risk of Wilm's tumour, hepatoblastoma and neuroblastoma. The majority of cases are sporadic (85%) and can be caused by a variety of genetic defects, most frequently abnormal regulation of gene transcription in the imprinted domain of chromosome 11p15.5. In some cases Beckwith-Wiedemann syndrome has been shown to be associated with genetic mutations on chromosome 11 with the IGF-2 (growth factor) and CDKN1C (inhibitor of cell proliferation) genes specifically implicated.

Management of omphalocele is dependent on its size and the presence of other anomalies. The prognosis of infants with omphalocele is mainly influenced by the severity of the associated anomalies as opposed to the abdominal wall defect itself. Indeed, the size of the abdominal wall defect does not correlate well with the incidence of associated anomalies. It has been shown that smaller defects have a higher risk of genetic and cardiac abnormalities. In our case management was conservative and at 6 months a small umbilical hernia was evident.

CrossMark

To cite: Smith AM, Healy DB, Ryan CA, et al. *BMJ Case Rep* Published online: [please include Day Month Year] doi:10.1136/bcr-2015-209695

Learning points

- ▶ Detection of omphalocele may not be straightforward in all cases.
- ▶ Small defects have higher risk of associated anomalies.
- ▶ Surgical assessment should be sought but surgical correction may not always be necessary.

Contributors Chart review and case report was performed and written by AMS and DBH. CAR was the attending consultant to the infant detailed in the case report.

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

Copyright 2015 BMJ Publishing Group. All rights reserved. For permission to reuse any of this content visit <http://group.bmj.com/group/rights-licensing/permissions>.
BMJ Case Report Fellows may re-use this article for personal use and teaching without any further permission.

Become a Fellow of BMJ Case Reports today and you can:

- ▶ Submit as many cases as you like
- ▶ Enjoy fast sympathetic peer review and rapid publication of accepted articles
- ▶ Access all the published articles
- ▶ Re-use any of the published material for personal use and teaching without further permission

For information on Institutional Fellowships contact consortiasales@bmjgroup.com

Visit casereports.bmj.com for more articles like this and to become a Fellow