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Original Article

Dandy–Walker syndrome

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الملخص

أهدف البحث: متلازمة داندي- والكر هي اعتلال نادر يتسم بعدم تكون كامل أو جزئي لدودة المخيخ، وتوسع كيسي للبطين المخي الرابع وتضخم الحفرة الدماغية الخلفية. ولا يعرف السبب الدقيق لحدوثه، على الرغم من وجود بعض التقارير التي ربطت بينه وبين عوامل الخطورة مثل العدوى الفيروسية للأمهات (الحصبة الألمانية، والتوكسوبلازما، والفيروس المضخم للخلايا) وتناول الكحول. تتراوح نسبة الإصابة بين واحد من كل 500,2 ولادة إلى واحد من كل 000,100 ولادة. قد يكون السبب في هذا الاختلاف الشاسع هو محدودية الحالات المنشورة، حيث أن معظم البيانات المتاحة هي تقارير أو سلسلة حالات متفرقة.

طرق البحث: أجريت هذه الدراسة الاستعادية على السجلات الطبية لحديثي الولادة المصابين بمتلازمة داندي- والكر الذين تم تنويمهم بوحدة الرعاية المركزة لحديثي الولادة بمستشفى جامعي في الفترة بين يناير 2001م وديسمبر 2010م.

النتائج: تنوم ثمانية رضع بمتلازمة داندي- والكر خلال فترة الدراسة، بمعدل عام 400/1 ولادة حية. وكانت نسبة الإناث إلى الذكور 7,1:1 (كان أحد الرضع ملتبس الجنس وتبين لاحقا أنه ذكر). كان متوسط عمر الحمل 39 أسبوعا (يتراوح بين 36-40 أسبوعا)، ومعدل الوزن عند الولادة 716,2 جرام (يتراوح بين 965,1 -335,3 جرام). تم تشخيص المتلازمة لنصف الرضع قبل الولادة. وكان لدى جميع الرضع استسقاء دماغي، وخمسة لديهم عيوب عصبية أخرى؛ أما عيوب خارج القحف فقد وجدت لدى 50% من الرضع. عاش جميع الرضع حتى خروجهم.

الاستنتاجات: على الرغم من أن العديد من نتائجنا تتفق مع البيانات المنشورة، فإن معدل متلازمة داندي- والكر في دراستنا كان أعلى بكثير من أي دراسة سابقة. وهذا يتطلب المزيد من البحوث لتفسير هذه النتائج غير المتوقعة.

الكلمات المفتاحية: متلازمة داندي-والكر; تضخم الرأس; تضخم البطين المخي

Abstract

Objectives: Dandy–Walker syndrome is a rare disorder characterised by complete or partial agenesis of the vermis, cystic dilatation of the fourth ventricle and an enlarged posterior fossa. The precise aetiology is unknown, although there have been reports of associations with risk factors like maternal virus infections (rubella, toxoplasma, and cytomegalovirus) and alcohol consumption. The reported incidence varies between one per 2500 births to one per 100,000 births. This huge difference may be due to the limited published case series, as most of the available data are sporadic case reports or series.

Methods: A retrospective review was conducted of medical records of neonates with Dandy–Walker syndrome admitted to the neonatal intensive care unit of a university hospital between January 2001 and December 2010.

Results: Eight infants with Dandy–Walker syndrome were admitted during the study period, giving an overall incidence of 1/400 live births. The female-to-male ratio was 1.7:1 (one infant had ambiguous gender but was later found to be male). The mean gestational age was 39.0 weeks (range, 36–40 weeks), and the mean birth weight was 2716 g (range, 1965–3335 g). The syndrome was diagnosed in half the infants prenatally. All infants had associated hydrocephalus, and five had other neurological anomalies; extra-cranial anomalies were seen in 50% of infants. All infants survived to discharge.

Conclusion: Although many of our results were consistent with published data, the incidence of Dandy–Walker syndrome in our study was much higher than any

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reported previously. Further research is required to elucidate this unexpected finding.

Keywords: Dandy–Walker syndrome; Macrocephaly; Ventriculomegaly

Introduction

Dandy–Walker syndrome is a rare condition, which consists of hypoplasia of the cerebellar vermis, dilatation of the fourth ventricle and an enlarged posterior fossa. The syndrome is considered to be the commonest cerebellar malformation. It is poorly understood, and the incidence is unknown. Although this malformation can be diagnosed prenatally by neuroradiology, identifying patients is difficult, as there is no specific symptom or sign. It is not uncommonly detected from the associated hydrocephalus. The outcome is highly variable and ranges from normal or nearly normal development to profound disability or even early death. Data on this disease in Saudi Arabia are limited.

This study involved examination of the records of all infants with Dandy–Walker syndrome seen at our centre in the past 10 years and an analysis of the clinical and epidemiological aspects.

Materials and Methods

This retrospective study was carried out at the neonatal intensive care unit of the largest teaching hospital in the region and the only Government hospital that provides level III neonatal services in the city. The intensive care unit has a capacity of 18 beds.

All cases of Dandy–Walker syndrome born at or referred to the centre between January 2001 and December 2010 were reviewed. The information obtained from the files included gestational age, sex, Apgar score, birth weight, head circumference, place and mode of delivery, nationality, length of hospital stay, associated morbidity and mortality rate.

Microsoft Excel spreadsheets were used for data collection, and the data were analysed with SPSS version 15.0. Means, ratios and percentages were calculated. The study was authorised by the ethical committee.

Results

During the study period, eight infants with Dandy–Walker syndrome were admitted to the neonatal intensive care unit (Table 1), giving an overall incidence of 1/400 live births. Seven infants were Saudis. Three were born vaginally and five by caesarean section. Two infants were boys, five were girls, and one infant was born with ambiguous genitalia but was later found to be male, giving a female-to-male ratio of 1.7:1. The mean gestational age was 39.0 weeks (range, 36–40 weeks); one infant was borderline premature, but all the other infants were born at term. The mean birth weight was 2716 g (range, 1965–3335 g). The mean head circumference was 35.6 cm (range, 31–43.5 cm). The mean Apgar scores were 6 and 7 at the 1st and 5th minute, respectively.

Dandy–Walker syndrome was diagnosed in four infants prenatally. The diagnosis in six infants was confirmed by computerised tomography scan and in two by magnetic resonance imaging. All the infants were discharged home after a mean hospital stay of 26.5 days (range, 7–58 days).

Table 1: Overall findings in eight infants born with Dandy–Walker syndrome.

	Patient 1	Patient 2	Patient 3	Patient 4	Patient 5	Patient 6	Patient 7	Patient 8
Sex	F	F	M	F	F	F	M[a]	M
Mode of delivery	Vaginal	Caesarean section	Vaginal	Vaginal	Caesarean section	Caesarean section	Caesarean section	Caesarean section
Gestational age (weeks)	39	39	39	39	40	41	39	36
Nationality	Saudi	Saudi	Algerian	Saudi	Saudi	Saudi	Saudi	Saudi
Birth weight (g)	1965	3080	2500	2840	3125	2090	2795	3335
Length (cm)	45.0	45.5	46.0	50.0	47.0	46.0	47.0	51.5
Head circumference (cm)	30.5	37.0	31.0	35.5	40.0	30.5	37.0	43.5
Apgar score 1st min	7	5	7	6	6	3	5	2
Apgar score 5th min	8	8	8	7	9	6	7	2
Diagnosed by	CT	CT	CT	CT	MRI	MRI	CT	CT
Hydrocephalus	Present	Present	Present	Present	Present	Present	Present	Present
Other cranial findings	Encephalocoele and lisscencephaly	None	Multiple cysts and skull defect	Joubert's syndrome	Hydrancephaly	Lisscencephaly	None	Corpus callosum agenesis
Extra-cranial	Intrauterine growth restriction. micrognathia, microtia and patent ductus arteriosus	None	None	None	Patent ductus arteriosus	None	Hypospadias, ventricular septal defect and patent ductus arteriosus	Patent ductus arteriosus
Hospital stay (days)	25	41	58	7	27	18	15	21

[a] Ambiguous genitalia.