Child's Nervous System (2019) 35:1429–1434
https://doi.org/10.1007/s00381-019-04164-6

CASE REPORT

Basilar invagination in a child with atlanto-occipital subluxation and suspected prenatal Dandy–Walker malformation

Isamu Miura[1] · Yasuo Aihara[1] · Tetsuryu Mitsuyama[1] · Kentaro Chiba[1] · Hiroshi Nakano[1] · Takakazu Kawamata[1]

Received: 24 March 2019 / Accepted: 16 April 2019 / Published online: 17 May 2019

Abstract

Background and purpose Although advances in imaging have allowed earlier and more accurate diagnosis of various fetal anomalies, Dandy–Walker malformation (DWM) remains one of the more challenging central nervous system anomalies to diagnose accurately before birth. Basilar invagination (BI), which is a dislocation of the dens in an upward direction, is occasionally accompanied by Klippel–Feil syndrome (KFS). We report a pediatric case of BI caused by atlanto-occipital subluxation (AOS) in KFS, suspected of having DWM prenatally but head magnetic resonance images (MRI) showed no evidence of that at 7 months of age.

Case At 28 weeks of gestation, fetal MRI study revealed a small cerebellar vermis, leading us to suspect a DWM. The patient was born at 40 weeks of gestation. Head CT showed inferior vermian hypoplasia without findings of hydrocephalus. Cervicothoracic CT showed cervical lamina assimilations, thoracic hemivertebrae, and cervicothoracic scoliosis. He was diagnosed with Dandy–Walker variant and KFS. At 7 months of age, head MRI showed near normal cerebellum and vermis and there was no evidence of the DWM. He did not have intellectual or developmental delay and imaging studies were performed periodically. At 9 years of age, an already existing cough headache deteriorated. Three-dimensional reconstructed images from CT scan showed C1 hypoplasia, fusion of C1 and C2, BI, and AOS. Sagittal T2-weighted MRI showed protrusion of cerebellar tonsils inferiorly to the level of the posterior arch of C2. Serum calcium, phosphate, and parathyroid hormone levels were normal. The diagnosis was tonsillar herniation related to BI following AOS in KFS. Posterior occipitocervical fixation was performed under traction.

Conclusions We found out two important clinical issues: DWM findings after birth can be disappearing and BI can present sequential deterioration because of AOS in KFS. Our observation indicated the possible prognosis of pediatric BI with long follow-up and can help us decide on its surgical treatment timing when associated with AOS.

Keywords Dandy–Walker malformation · Basilar invagination · Klippel–Feil syndrome · Atlanto-occipital subluxation

Introduction

Dandy–Walker malformation (DWM) consists of a large posterior fossa, splayed cerebellar hemispheres, and a large vermian defect [1]. If posterior fossa is not large, it is called Dandy–Walker variant (DWV) [2]. Although advances in imaging have allowed earlier and more accurate diagnosis of various fetal anomalies, DWM remains one of the more challenging central nervous system anomalies to diagnose accurately before birth [3]. By the way, Klippel–Feil syndrome (KFS) is a congenital union of cervical vertebrae caused by failure of segmentation during embryonal development and frequently accompanies conditions such as cranial malformations, atlas assimilation, or dens anomalies [4]. Basilar invagination (BI), which is a dislocation of the dens in an upward direction, is occasionally accompanied by KFS [4, 5]. We report a pediatric case of BI caused by atlanto-occipital subluxation (AOS) in KFS, who was suspected to have DWM prenatally but head magnetic resonance images (MRI) showed no evidence of the DWM at 7 months of age.

✉ Yasuo Aihara
yaihara@twmu.ac.jp

[1] Department of Neurosurgery, Tokyo Women's Medical University, 8-1 Kawada-cho Shinjuku-ku, Tokyo 162-8666, Japan

Case report

History and examination

A fetus at 24 gestational weeks was observed to have a small cerebellar vermis by fetal ultrasonographic examination. There were no findings of hydrocephalus or myelomeningocele.

At 28 weeks of gestation, fetal MRI study confirmed the small cerebellar vermis in a large posterior fossa cyst with tentorium displaced upwards, leading us to suspect a DWM (Fig. 1). The patient was born at 40 weeks of gestation in an uneventful delivery with a birth weight of 2678 g. The 1- and 5-min Apgar scores were 8 and 9, respectively, and he had no detectable neurological complications. Chromosome test was normal. At 2 days of age, head CT showed inferior vermian hypoplasia without imaging findings of hydrocephalus (Fig. 2). Cervicothoracic CT showed cervical lamina assimilations, thoracic hemivertebrae, and cervicothoracic scoliosis. He was diagnosed with DWV and KFS. There were no imaging findings of BI or AOS. At 7 months of age, head MRI showed near normal cerebellum and vermis, in particular, and there was no evidence of the DWM (Fig. 3).

He did not have intellectual or developmental delay and imaging studies were performed periodically. At 8 years of age, he started having a cough headache. A year later, his headache became stronger but he did not have sleep apnea or neurological deficit. He could flex and extend his neck but was not able to rotate it. The plain cervical X-rays, lateral view, showed BI (Fig. 4). Dynamic imaging demonstrated slight occipitocervical instability. Three-dimensional reconstructed images from a CT scan showed assimilations of C1 posterior arch, lateral mass, and C2 lamina, a fusion of C1 anterior arch and dens and AOS (Fig. 4). A sagittal CT reconstruction image showed that the tip of the odontoid process was 12 mm above Chamberlain's line and clivoaxial angle was 122°. The angle between the floor of the anterior fossa and clivus (basal angle) measured 130°, which is below the top normal limit of 143°. Sagittal T2-weighted MRI showed the cerebellar tonsils protruding inferiorly to the level of the posterior arch of C2. Serum calcium, phosphate, and parathyroid hormone levels were normal. The diagnosis was tonsillar herniation in BI following AOS in KFS. Posterior occipitocervical fixation was performed under traction.

Operation and postoperative course

The patient was placed in a prone position under general anesthesia with fibroscopic endotracheal intubation. We performed skull traction under SEP monitoring. Lateral radiographs taken during traction showed partial cranio-caudal reduction.

We performed posterior occipitocervical fixation using a plate-rod system. A posterior midline longitudinal skin incision was made from the external occipital protuberance to the spinous process of C4. Decompression of the foramen magnum was performed. The spine was exposed bilaterally to the lateral margin of the facet joints. The fixation constructs employed the VERTEX MAX System (Medtronic, Sofamor Danek, Memphis, TN, USA). Occipital plates were fixed to the midline of the occiput by three 4.5-mm and one 5-mm screws. Two translaminar screws (3.5 × 20–22 mm) were inserted into the C2 laminas. Titanium rods were bent and cut to approximate the occipitocervical curvature and were attached to each screw head, according to the construct design. After decortication of C2 lamina, a bone graft harvested from the ilium was transplanted between the occipital bone and C2 lamina (Fig. 5). Intra-operative spinal monitoring did not indicate abnormalities.

Postoperatively, there was no neurological deficit. An Ortho Collar (Arizono Bespoke, Japan) was applied after surgery. Recovery was satisfactory and the patient was discharged on the 14th postoperative day. His cough headache disappeared. The clivoaxial angle increased from 122° preoperatively to 131° postoperatively (Fig. 5).

Discussion

We had two important clinical observations in our case. The first was that DWM findings after birth can disappear subsequently and the second was that BI can show gradual

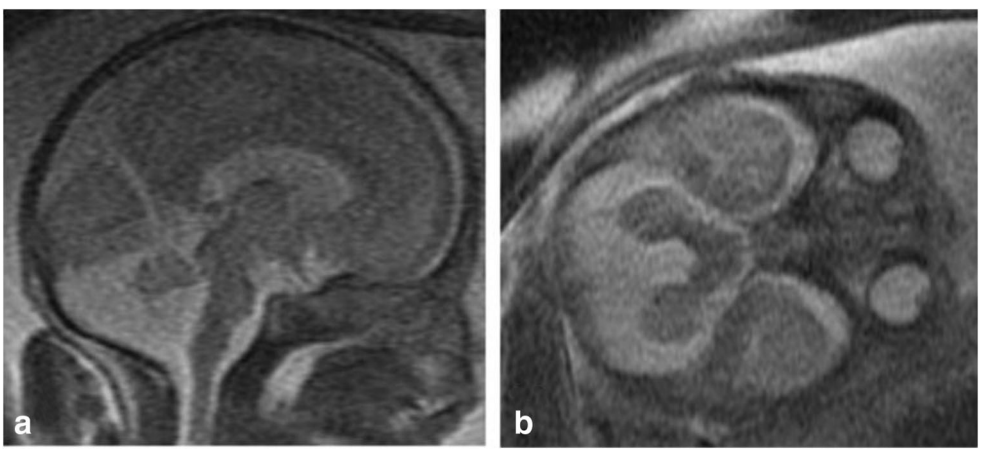

Fig. 1 Fetal MRI, T2-weighted image, sagittal view (**a**) and axial view (**b**), showing a cystic forth ventricle. The posterior fossa has a slightly larger size and the tentorium has a steeper slope

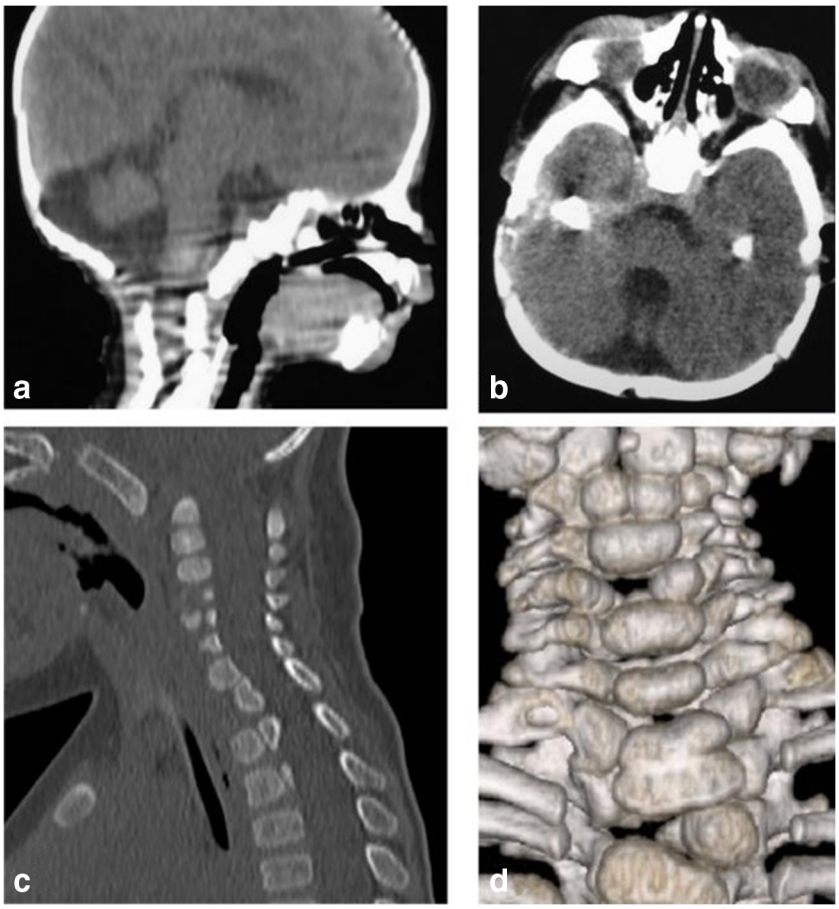

Fig. 2 Head CT, a sagittal CT reconstruction image (**a**) and axial view (**b**), showing a cystic fourth ventricle. The posterior fossa does not have a large size and there are no findings of hydrocephalus. Cervical CT, a sagittal CT reconstruction image (**c**) and three-dimensional reconstructed images (**d**), showing cervical lamina assimilation, thoracic hemivertebrae, and scoliosis. There are no imaging findings of basilar invagination

deterioration because of AOS in KFS without generalized skeletal diseases, as hyperparathyroidism, Paget disease, or osteogenesis imperfecta.

DWM is a rare congenital malformation of the posterior fossa with an incidence ranging between 1/25,000 and 1/35,000 births [1]. Numerous malformations are mentioned in the literature in association with both DWM and DW complex. They include partial or total corpus callosum agenesis or hypoplasia and occipital meningoceles [1, 6]. In our case, fetal MRI study revealed a small cerebellar vermis, a large posterior fossa cyst, and a tentorium displaced upwards, leading us to suspect a DWM. After birth, head CT showed no large posterior fossa so he was diagnosed with DWV. Approximately 70–90% of patients in DWM have hydrocephalus [2]. However, the size of the ventricles in our case was normal at birth and as hydrocephalus develops in more than 80% of DWM patients mostly in the first 3 months of life [1]; we insisted on a close follow-up. At 7 months of age, head MRI showed near normal

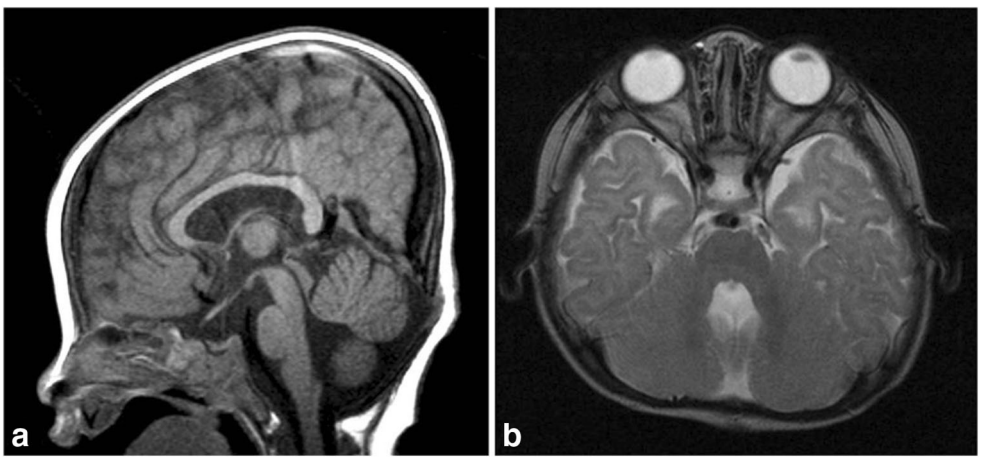

Fig. 3 Sagittal on the FLAIR images (**a**) and axial on the T2-weighted images (**b**) views of the head MRI showing intact cerebellum and vermis and there was no evidence of the DWM at 7 months of age

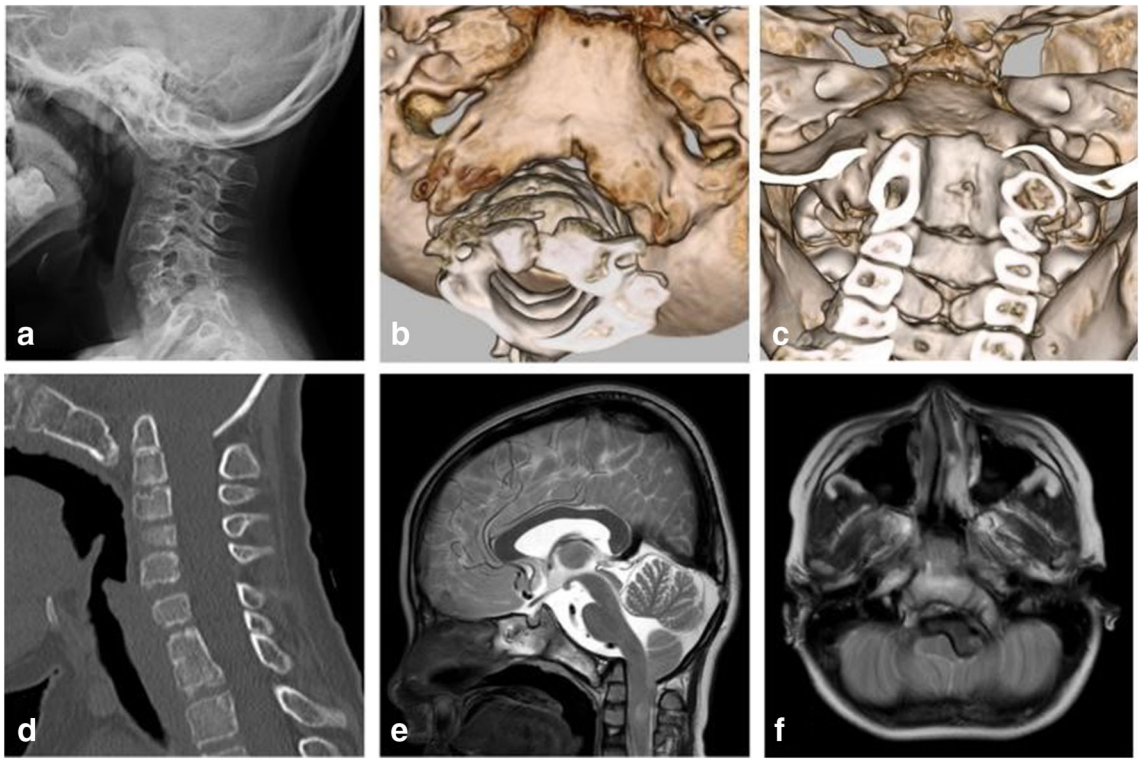

Fig. 4 Lateral cervical radiography (**a**) showing basilar invagination. A coronal view (**b**) and view from caudal (**c**) of three-dimensional reconstructed images from CT scan, showing hypoplasia of the condyles and atlanto-occipital subluxation. A sagittal CT reconstruction image (**d**), showing that the tip of the odontoid process was 12 mm above Chamberlain's line and clivoaxial angle was 122°. Sagittal (**e**) and axial (**f**) views of the head magnetic resonance images (MRI), showing the cerebellar tonsils protrusion inferiorly to the level of the posterior arch of C2 and compression of the medulla oblongata

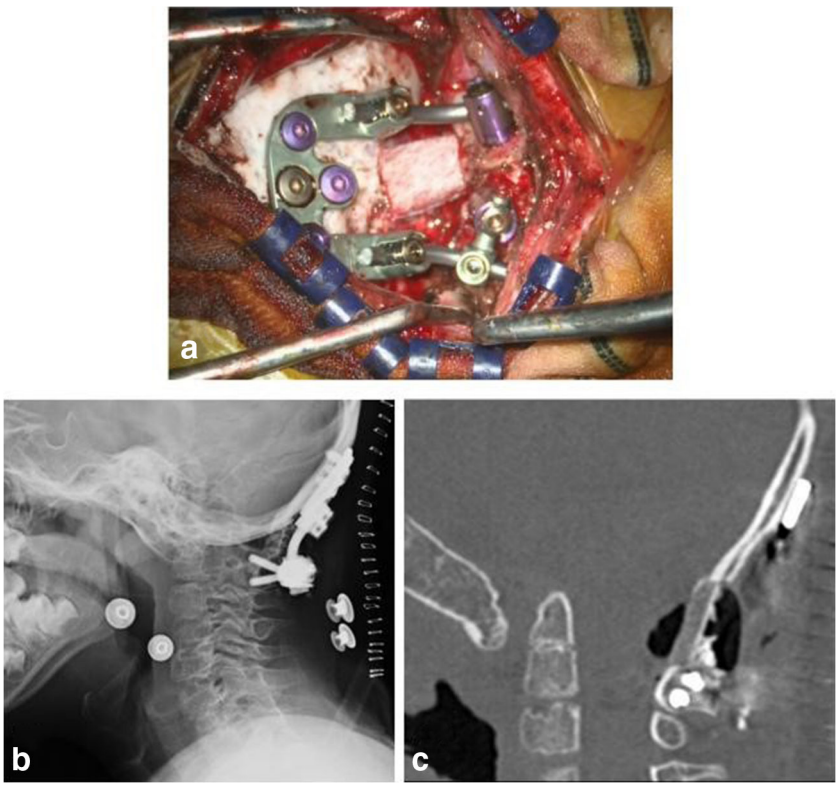

Fig. 5 Surgical view (**a**). Occipital plates were applied to the midline of the occiput and a rod was connected to the lamina screws. Postoperative lateral cervical radiography (**b**) demonstrating instrumented occipito-C2 fixation. A sagittal CT reconstruction image (**c**) after surgery demonstrating a solid bone graft has been transplanted between the occipital bone and C2

cerebellum, vermis, and no evidence of the DWM. In the fetus, diagnosis of DWM can be difficult [7]. There is only one report of disappearing DWM findings after birth [3]. Tsao et al. reported a myelomeningocele associated with an Arnold–Chiari malformation that had the sonographic findings of a DWM prenatally. The findings of DWM disappeared after birth. They assumed that the suspected DWM and hydromyelia seen on the prenatal sonogram were not caused by a morphologic abnormality of the vermis (which would not appear and disappear) but by dilatation of the fourth ventricle and herniation into the cervical spine. In our case, at 2 days of age, head CT showed inferior vermian hypoplasia, suspecting morphologic abnormality. Cerebellar vermis finishes its development from superior to inferior at about 17–18 gestational weeks [7]. At 7 months of age, head MRI showed almost normal cerebellum and vermis so we cannot deny the possibility that the suspected DWM seen on the prenatal sonogram and MRI was not caused by a morphologic abnormality but dilatation of the fourth ventricle.

The patient had also cervicothoracic spinal malformation including cervical lamina assimilation, thoracic hemivertebrae, and scoliosis and was diagnosed with KFS. There were no imaging findings of BI or AOS at birth. C1 was hypoplastic and fused with C2. Atlanto-occipital joints were considered also hypoplastic. With time, AOS deteriorated and BI progressed (Fig. 6) probably with an increase in child's activity (standing and walking) and additional axial, flexion, and extension load on the craniocervical junction. Secondary BI often occurs as a result of generalized skeletal diseases, including hyperparathyroidism, Paget disease, and osteogenesis imperfecta. However, our patient did not have clinical and laboratory findings suggesting these diseases.

Surgical treatment for a case as ours might be considered controversial. Continued follow-up with a cervical collar can be an option. However, the imaging studies of AOS and BI showed sequential deterioration. Cough headache was considered a symptom of tonsillar herniation coming from the relatively small posterior fossa with BI and the AOS, interfering with CSF dynamics. The possibility of future deterioration was considered highly probable because BI and AOS are on the background of a cervical malformation and dynamic imaging demonstrated slight atlanto-occipital instability. That required proper timing of the surgical treatment to prevent further neurological deficit although surgery is often challenging, especially in children.

Anterior decompression has been recommended due to the ventral compression of the spinal cord and medulla oblongata [8]. In our case, the AOS is the main pathology leading to the ventral compression; therefore, reduction and stabilization of the atlanto-occipital joints can be, to some extent, decompressive and be an effective treatment. The role of skull traction in the management of craniovertebral abnormalities has also been described [9]. Simsek et al. reported successful correction of BI by posterior decompression and fusion after 4 weeks of halo traction [9]. A halo traction, however, could have been a big burden on a 9-year-old patient. We performed direct traction on the surgical bed intraoperatively under general anesthesia. Recent reports also have demonstrated that instrumented internal occipitocervical fixation is a reasonable therapeutic option, even for pediatric patients [10]. Occipitocervical fixation by the combination of cervical screws and occipitocervical rod systems provided a high fusion rate [11]. We performed posterior occipitocervical fixation using the VERTEX MAX system (Medtronic, Sofamor Danek, Memphis, TN, USA) occipitocervical plate-rod systems. There were no complications and postoperative course was uneventful.

In conclusion, DWM findings after birth can be disappearing. They can be caused by dilatation of the fourth ventricle. On the same 9-year-old patient, we performed surgical treatment because of BI caused by AOS. We can estimate the prognosis of BI in the long term and time the surgical treatment at the earliest indication of neurological progression. The relationship of DWM findings and spinal malformation is not clear. Further observation and analysis of such cases might be helpful in understanding the relationships of the different elements of this condition.

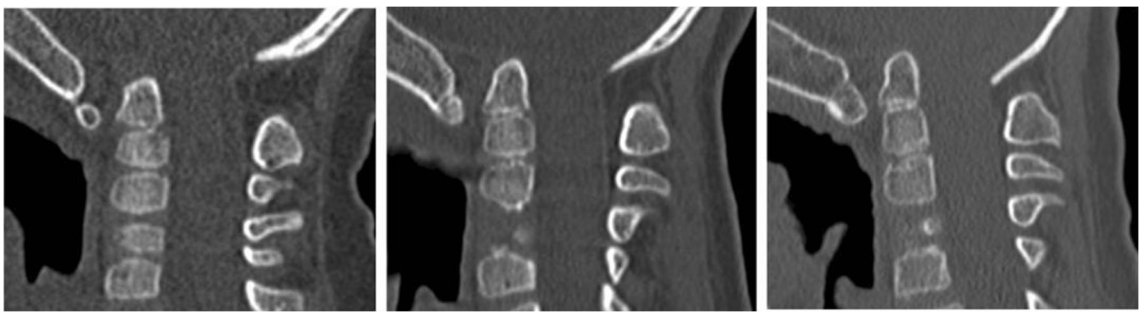

Fig. 6 Time course of a sagittal CT reconstruction image, at birth (**a**), at 5 years of age (**b**), and at 9 years of age (**c**), showing sequential deterioration of basilar invagination. Clivoaxial angle is respectively 131°, 126°, and 122°

Acknowledgments We thank Motoo Kubota, Department of Spinal Surgery, Kameda Medical Center, for his guidance and we gratefully acknowledge the radiological technologists, nurses, and staff of the Departments of Neurosurgery, Tokyo Women's Medical University in preparing this paper.

Declaration of funding source This study was supported by research funds of the Department of Neurosurgery, Tokyo Women's Medical University.

Compliance with ethical standards

Conflict of interest The authors report no conflict of interest concerning the materials or methods used in this study or the findings specified in this paper.

References

1. Hirsch JF, Pierre-Kahn A, Renier D, Sainte-Rose C, Hoppe-Hirsch E (1984) The Dandy-Walker malformation. A review of 40 cases. J Neurosurg 61:515–522
2. Klein O, Pierre-Kahn A, Boddaert N, Parisot D, Brunelle F (2003) Dandy-Walker malformation: prenatal diagnosis and prognosis. Childs Nerv Syst 19:484–489
3. Tsao K, Chuang NA, Filly RA, Barkovich AJ, Goldstein RB (2002) Entrapped fourth ventricle: another pitfall in the prenatal diagnosis of Dandy-Walker malformations. J Ultrasound Med 21:91–96
4. Ogihara N, Takahashi J, Hirabayashi H, Mukaiyama K, Kato H (2013) Surgical treatment of Klippel-Feil syndrome with basilar invagination. Eur Spine J 22(Suppl 3):S380–S387
5. Dokai T, Nagashima H, Nanjo Y, Tanida A, Teshima R (2011) Posterior occipitocervical fixation under skull-femoral traction for the treatment of basilar impression in a child with Klippel-Feil syndrome. J Bone Joint Surg (Br) 93:1571–1574
6. Hart MN, Malamud N, Ellis WG (1972) The Dandy-Walker syndrome. A clinicopathological study based on 28 cases. Neurology 22:771–780
7. Ecker JL, Shipp TD, Bromley B, Benacerraf B (2000) The sonographic diagnosis of Dandy-Walker and Dandy-Walker variant: associated findings and outcomes. Prenat Diagn 20:328–332
8. Menezes AH, VanGilder JC (1988) Transoral-transpharyngeal approach to the anterior craniocervical junction. Ten-year experience with 72 patients. J Neurosurg 69:895–903
9. Simsek S, Yigitkanli K, Belen D, Bavbek M (2006) Halo traction in basilar invagination: technical case report. Surg Neurol 66:311–314 discussion 314
10. Miyoshi Y, Yasuhara T, Date I (2011) Noonan syndrome with occipito-atlantal dislocation and upper cervical cord compression due to C1 dysplasia and basilar invagination. Neurol Med Chir (Tokyo) 51:463–466
11. Abumi K, Takada T, Shono Y, Kaneda K, Fujiya M (1999) Posterior occipitocervical reconstruction using cervical pedicle screws and plate-rod systems. Spine (Phila Pa 1976) 24:1425–1434