Case Report

Prenatally diagnosed type II Arnold Chiari Malformations-a rare congenital anomaly with some unusual associated defects

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Abstract

Arnold Chiari malformation (ACM) is one of the common anomalies of the craniovertebral junction involving both the skeletal as well as the neural structures. Among the four types of ACM, type II ACM is considered as commonest. A case of Type II Arnold Chiari malformations; a rare congenital anomaly with some unusual associated defects has been reported here. A 29 year old lady with 21weeks gestation was diagnosed to have a male foetus with type II ACM at antenatal clinic of department of Gynaecology, Sree Mookambika Institute of Medical Sciences, Kulasekaram, Tamil Nadu, India during routine antenatal check-up. Though the commonly observed malformations of type II ACM such as herniation of cerebellar tonsil, spina bifida and hydrocephalous were observed in the present case, it differed from general pattern of type II ACM in presence of a large cyst abutting spina bifida and mild scoliosis.

Keywords: Congenital Anomalies, Spina Bifida, Hydrocephalus, Scoliosis, Cerebellar Tonsil Herniation,

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Introduction

In 1891, Hans Chiari; an Austrian pathologist observed a set of complex congenital malformations associated with hindbrain disorders in paediatric autopsy specimens and termed the condition as Arnold Chiari malformations (ACM) [1, 2, 3]. It has been reported that incidence is gradually rising; may be because of increased detection with MRI, ultrasound and other radiological techniques. We report a very rare case of type II Arnold Chiari malformation which was detected prenatally during routine antenatal check-up.

Clinical observations and case presentation

A 29 year old lady came to the antenatal clinic of department of Gynaecology at our hospital for routine check-up of her pregnancy about 6 weeks after her last menstruation. During this visit to the hospital, her routine urine, blood and ultrasonographic tests were performed. Urine examination report was normal with some traces of albumin, 6-8 pus cells, and 15-20 epithelial cells. Her thyroid function test results and blood test reports were also normal. Ultrasonography performed at LMP 6 weeks showed a single intrauterine gestation with GS (9.5mm) corresponding to approximately less than 4 weeks. A corpus luteal cyst was also observed in left ovary. Fetal parts and cardiac activity were not visualised. Bilateral ovaries were normal in their echo-structure however; there was evidence of 12 mm cyst in the left ovary. The patient was suggested review scan after 2 weeks for assessment of fetal viability.

After 4 weeks, she made her next visit to our antenatal clinic for further follow-up. Ultrasonography was performed and it showed following fetal parameters. Biperietal diameter- 18 mm measurement corresponding to 12 weeks and 5 days, head circumference- 68 mm measurement corresponding to 12 weeks and 5 days and abdominal circumference- 66 mm measurement corresponding to 13 weeks and 2 days. Fetal cardiac activity was 162 beats per minute, amniotic fluid was adequate, placenta was posteriorly placed, internal os was closed and cervical length was

normal. An impression of single, live intrauterine gestation corresponding to approximately 12 weeks and 6 days was made and the lady was asked to come back to the ante-natal clinic after about 2 months for further follow-up.

After about 2 months, she was again subjected to ultrasonogrpahy [Figure 1] at about 21 weeks of gestation and following fetal parameters were observed. Biperietal diameter- 45mm; measurement corresponding to 19 weeks and 5 days, head circumference- 174mm; measurement corresponding to 20 weeks, abdominal circumference- 150 mm; measurement corresponding to 20 weeks and 2 days and femur length- 31mm; measurement corresponding to 19 weeks and 6 days. Fetal cardiac activity was 135 beats per minute, amniotic fluid was adequate, placenta was posterior left lateral and anterior upper body, internal os was closed and cervical length was normal. Estimated fetal weight was approximately 329 g. When foetus was scanned for anomaly, following observations were made.

1. Small posterior cranial fossa with mild herniation of cerebellum through the foramen magnum.
2. Mildly dilated lateral ventricles
3. Spina bifida in the region of upper lumbar spine.
4. A cystic structure at the posterior aspect of lumbar spine was also observed abutting spina bifida.
5. There appeared a communication between cystic structure and CSF of spinal canal indicating the possibility of meningocoele.
6. Scoliotic deformity was also noted in the lower thoracic and upper lumbar spine.

Keeping above observation in mind, a prenatal diagnosis of type II Arnold Chiari malformation is made. However, other routine blood urine and thyroid tests conducted during this visit were normal.

Consent for medical termination of pregnancy was taken after counselling the couple at about 21 weeks of gestation. Labour was induced and a male baby weighing about 400g was delivered vaginally. The foetus had mild hydrocephalous. Other gross observation of foetus confirmed our ultrasonographic findings such as spina bifida in the region of upper lumbar spine and cystic structure at the posterior aspect of lumbar spine abutting spina bifida. The cyst was about 3cm in diameter almost surrounding the region of spina bifida. The swelling was marked on ether siders and above the spina bifida. (Figures2, 3 and 4) The scoliosis was confirmed by palpation. The umbilical cord had normal attachment on the placenta. The gross observation of placenta revealed a bilobed placenta. (Figures 2 and 3) Four placental bits were sectioned and stained with haematoxylin and eosin. Microscopic observation showed normal chorionic villi, decidual tissue and membranes without any abnormality (Figure 5). Mother was discharged on folic acid and iron supplements.

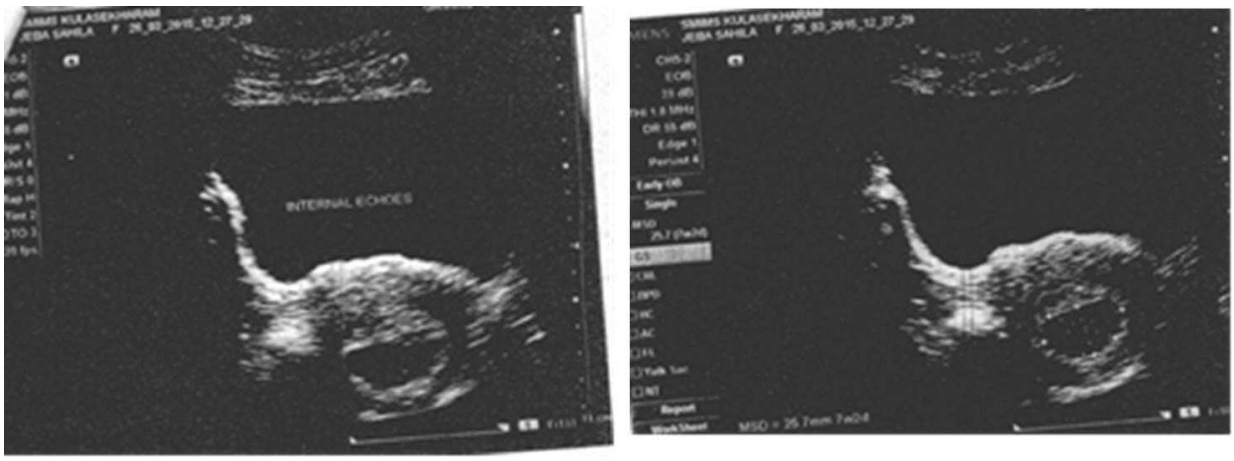

Figure 1: Ultrasonographic images taken at about 21 weeks of gestation

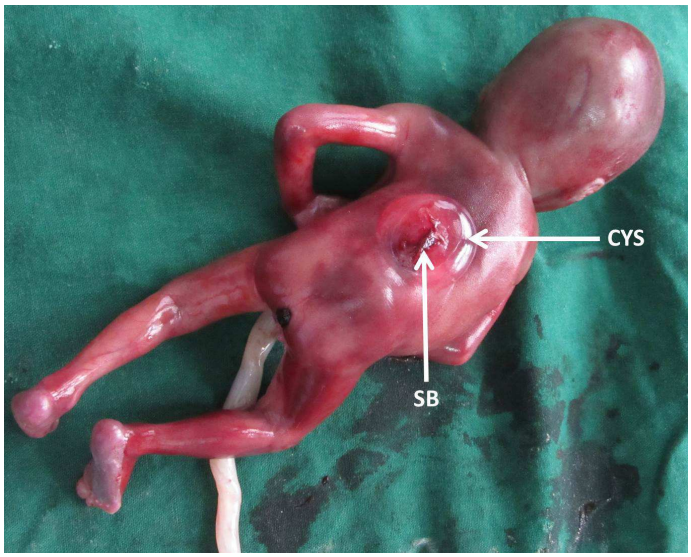

Figure 2: Aborted foetus showing spina bifida in the region of upper lumbar spine (SB) and a cystic structure measuring about 3cm at the posterior aspect of lumbar spine abutting spina bifida (CYS).

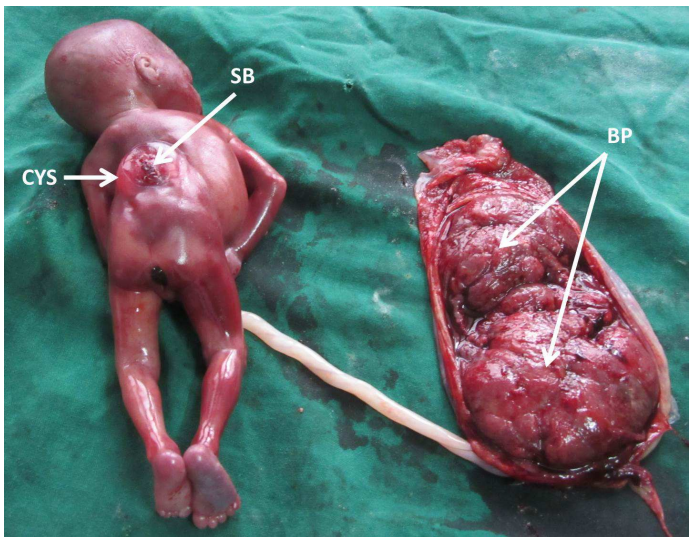

Figure 3: Aborted foetus with its bilobed (BP) placenta showing spina bifida in the region of upper lumbar spine (SB) and a cystic structure measuring about 3cm in diameter at the posterior aspect of lumbar spine abutting spina bifida (CYS).

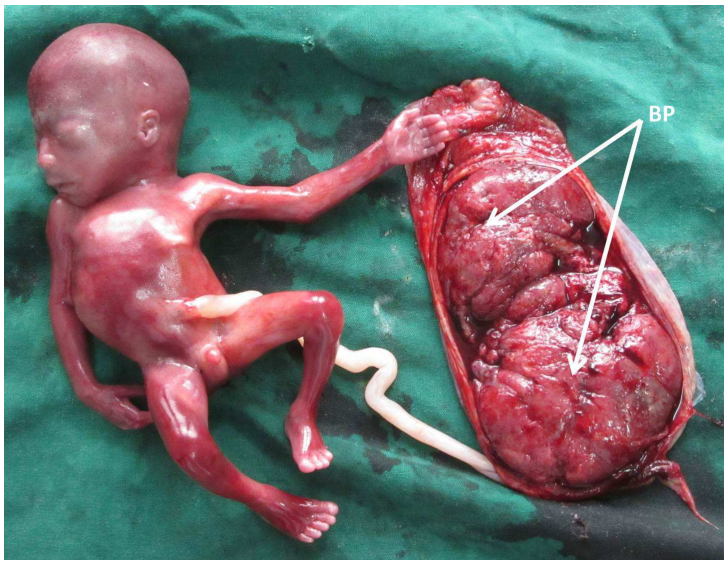

Figure 4: Aborted foetus with its bilobed (BP) placenta. Mild hydrocephalous and scoliosis can be noted.

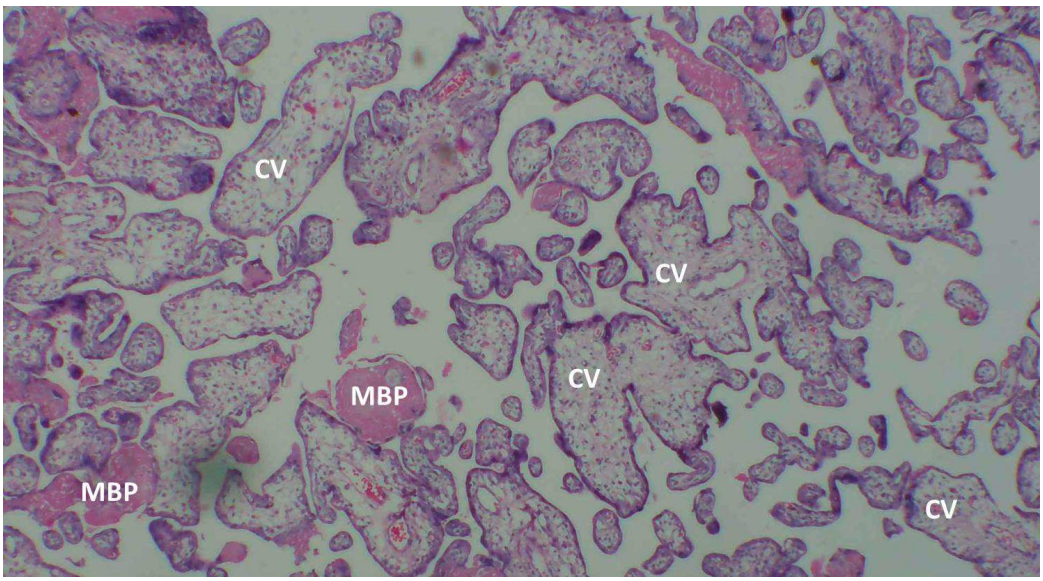

Figure 5: Photomicrograph of the haematoxylin and eosin stained section through the placenta showing normal structure of chorionic villi (CV). MBP- Meternal blood pool in the intervillous space

Discussion

Generally, Arnold Chiari malformations are classified into four types. Though, originally Chiari classified these set of CNS malformations into only three types; Types I, II and III, later a IV type was added. [1, 4] In addition to the traditional classification, detailed evaluations of such cases using various imaging techniques have introduced more refined forms of the malformation scores currently named as Chiari 0 to Chiari 1.5. Also in practice are other terms like "asymptomatic" and "incidental" ACM depending upon the extent of cerebellar herniation and other clinical findings [4]. Though the present case broadly falls under Type II ACM with Chari score 1, presence of

other anomalies associated with it makes the case complicated and unique.

Type II ACM is generally considered as commonest of the four types. In Chari type II malformation, rounded projections from cerebellar tonsils are displaced inferiorly into the cervical part of the vertebral canal through the foramen magnum. Generally, there is little or no damage to the cerebellar vermis, the fourth ventricle and medulla oblongata. There are reports of mild hydrocephalous in about 25% cases, syringomyelia in about 60% of cases, skeletal anomalies in about 25% of cases, basilar invagination in about 50% of cases and Klippel Feil syndrome in about 10% of cases [3, 5]. There are also reports of neural tube defects, calvarial defects, fenestrated falx, hypoplastic tentorium, wider foramen magnum, medullary kink and corpus callosal hypoplasia. [5, 6] It can be noted that in the present case, some of these defects were observed; there were other defects such as presence of a large cyst abutting spina bifida, dilated lateral ventricles and mild scoliosis are seldom associated with type II ACM.

There are number of reports about different approaches of treatment for type I ACM in both paediatric and adult patients. [7, 8, 9]. As far as treatment for type II ACM is concerned, very few reports are available. One of the most commonly performed treatment procedure is atlantoaxial stabilization [9]. In addition, there is a report that type II ACM can be prevented by preconceptional folic acid and Vitamin B 12 supplementation [5]. Complexity of disabilities involving wide range of neural and skeletal malformations, prenatal diagnosis of the malformation and possibility of medical termination of pregnancy as in the present case may be are some of the reasons for lack of abundancy about literature on treatments for type II ACM.

Conclusion

Arnold Chiari malformation is one of the common anomalies of the craniovertebral junction involving both the skeletal as well as the neural structures. Among the four types of ACM, type II ACM observed in this case is considered as commonest. Hoverer, present case differs from general pattern of type II ACM in presence of a large cyst abutting spina bifida and mild scoliosis.

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