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Arnold Chiari Malformation Type II with Club Hand Deformity.

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ABSTRACT

Arnold-Chiari malformation is a not uncommon congenital abnormality of CNS, characterized by downward displacement the parts of the cerebellum, fourth ventricle, pons and medulla oblongata into the spinal canal, likely a result of a small posterior fossa. This malformation is one of causative factor of death in neonates and infants. A detailed understanding of the direct and indirect sonographic findings is necessary for diagnosis of Chiari II malformation in the developing fetus. In this case report, we present a Chiari malformation II detected at 19 weeks of gestation detected during roution sonography study. The Role of prenatal sonography in recognition of the malformation, uncommon association of upper limb anomaly of club hand deformity is discussed.

Keywords: Arnold Chiari Malformation Type II, Lumbosacral Meningocele, Club foot, Club hand

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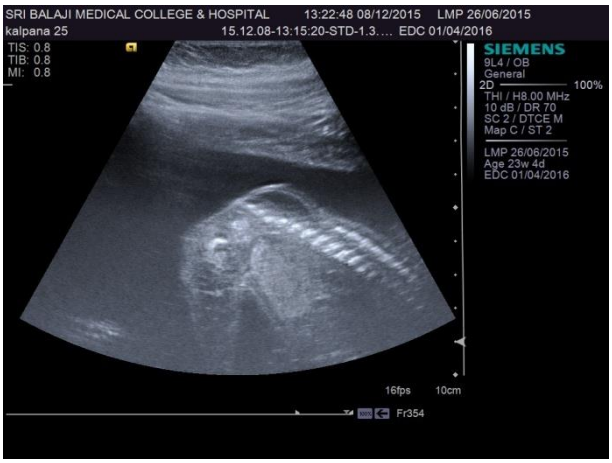
INTRODUCTION

Chiari II malformations are encountered relatively commonly with an incidence of ~1:1000 live births. When a child is born with a myelomeningocele the vast majority (~95%) have an associated Chiari II malformation [1]. The Arnold-Chiari malformation is a congenital deformity characterized by displacement of parts of the cerebellum, fourth ventricle, pons and medulla oblongata into the spinal canal.The exact cause of Chiari malformation is unknown; it is thought that a problem during fetal development may cause the abnormal brain formation. Chiari malformation may be caused by exposure to harmful substances during fetal development or associated with genetic problems or syndromes that may have a tendency to run in families. Theories suggest that the following may predispose the fetus to problems that affect the normal development of the head during pregnancy: exposure to hazardous chemicals/substances,lack of proper vitamins and nutrients in the diet, infection, prescription or illegal drug and alcohol consumption.

Type I consists of inferior displacement of the tonsils and cerebellum without displacement of the fourth ventricle or medulla. ArnoldChiari malformation type II is the most common and seen in neonates and infants which characterized by displacement of cerebellar tonsils, parts of the cerebella fourth ventricle, pons and medulla oblongata through the foramen magnum into the spinal canal .This is usually associated with hydrocephalus and myelomeningocele (2) (Fig 1, 2). Chiari III malformation is a high cervical encephalomeningocele in which the medulla, fourth ventricle, and virtually the entire cerebellum reside (3). The obvious sonographic findings are the frontal bone scalloping (lemon sing) (fig 5 & 6) and absent cerebellum or abnormal anterior curvature of the cerebellar hemispheres (banana sign) (4)fig 5 & 6. We report one case of Chiari malformation II detected at 19 weeks of gestation in routinely sonographic monitoring and describe direct and indirect sonographic findings that are the basis for diagnosis of Chiari II malformation and uncommon association of upper limb deformity such as club hand.



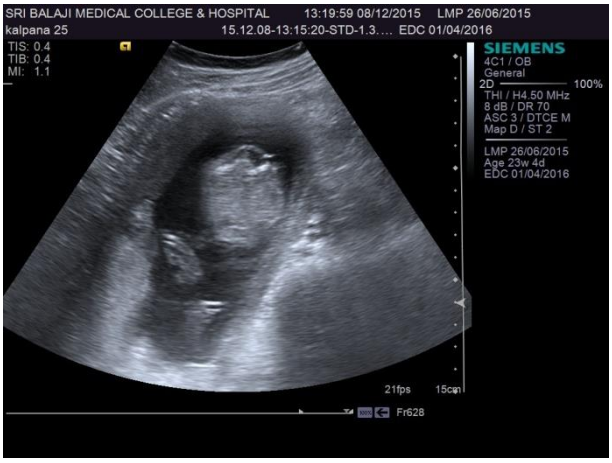


Fig 1&2shows myelomeningocele in lumbosacral region.

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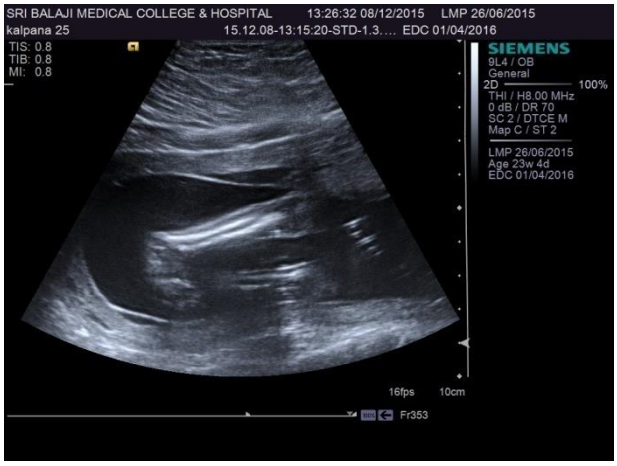


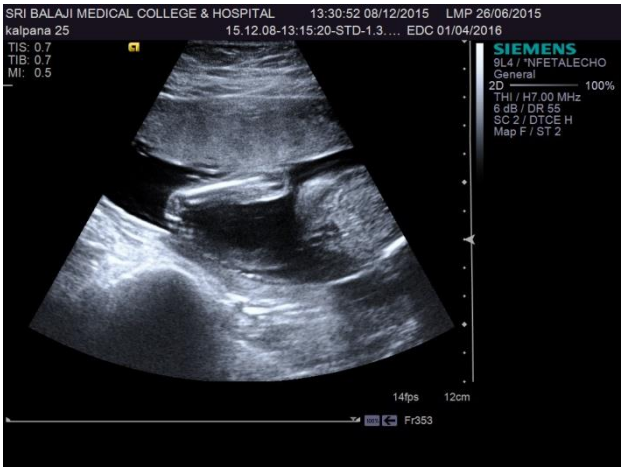
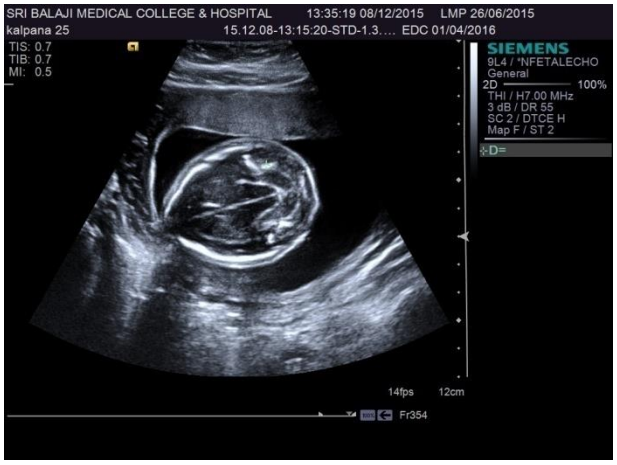


Fig3&4show club foot and club hand deformity



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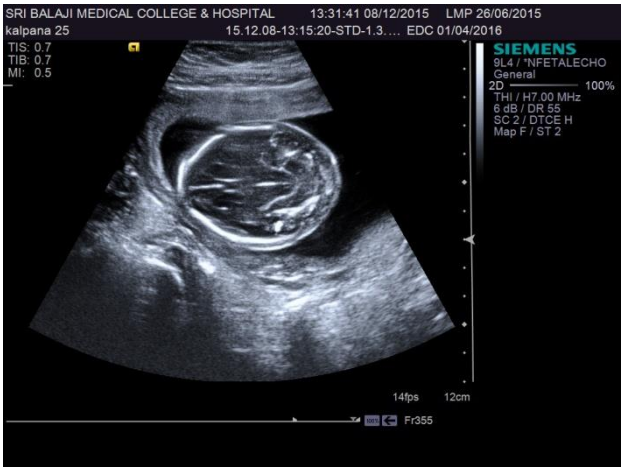

Fig 5&6 show lemon shaped skull, banana shaped cerebellum and non visualization of cistern magna.

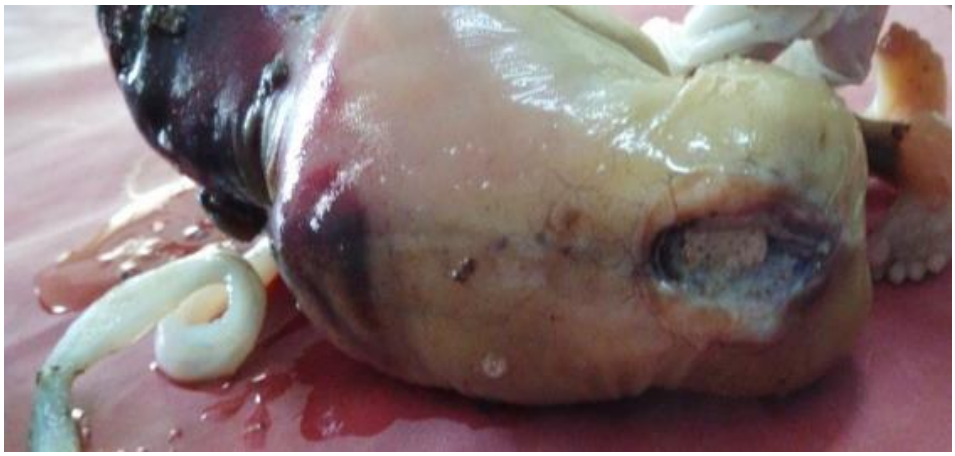

Image 1shows lumbosacralmeningocele with skin defect.

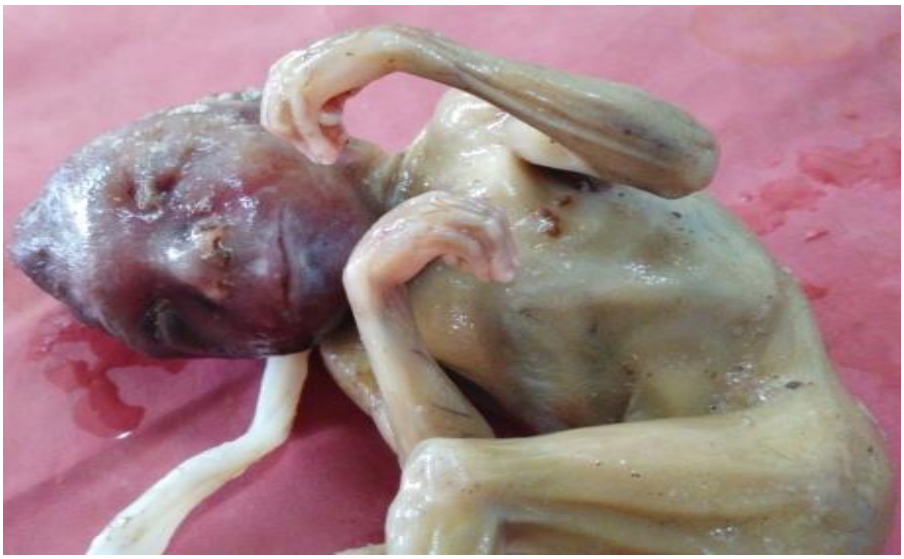

Image 2 showsDeformed skull and bilateral club hands

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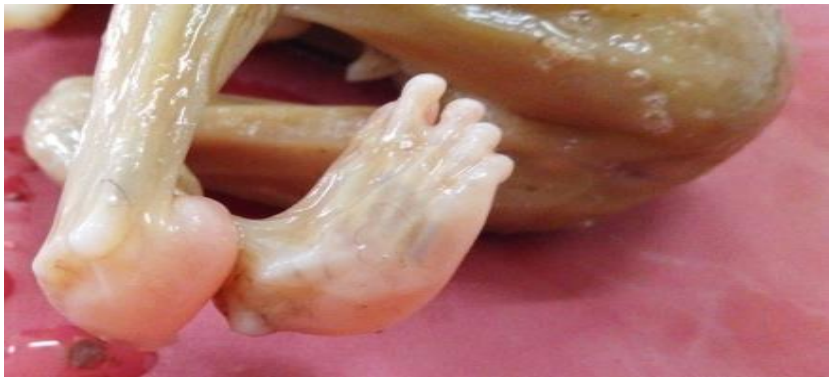

Image 3 shows bilateral club foot

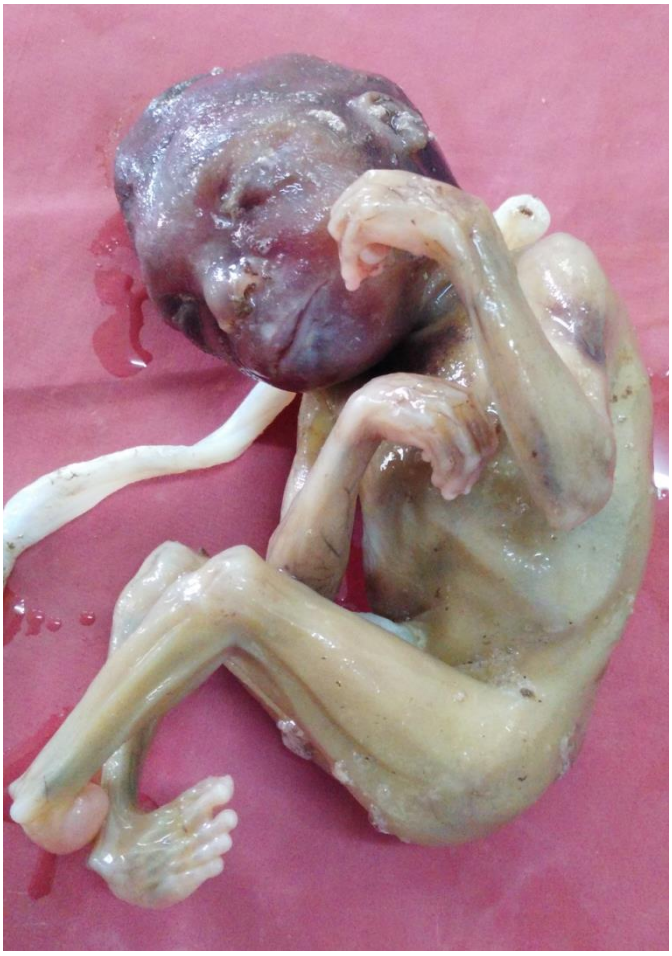

Image 4 shows full view of the fetus with bilateral club hand and club foot.

Case Report

A 25-year-old pregnant woman, gravida 2, para 0, abortion 1, with history of second degree consaguinaneous marriage and history of bipolar disorder on regular treatment and no history of familial genetic disorders and came to Department of radio diagnosis, SreeBalaji medical college, Chennai, for anomaly scan at 20 weeks with history of amenorrhea. Patient was a known case of Bipolar disorder since one year, was on (Antipsychotics) treatment for the same and on regular treatment, till her pregnancy was confirmed. Not a known case of diabetes mellitus, hypertension, tuberculosis, bronchial asthma. No previous history of surgery.

First pregnancy ended in spontaneous incomplete abortion and followed by treated with dilatation and curettage. Current pregnancy, first trimester scan was done elsewhere; NT was normal and measures 1.0mm.Patient came here for anomaly scan. In sonographic study at 20 weeks of gestation, multiple fetal anomalies including Lumbosacral meningomyelocele (Fig 1& 2), lemon sign (Fig 5 & 6), Banana sign and obliteration of cisterna magna (Fig 5 & 6) and Bilateral club foot deformity and bilateral club hand deformity (Fig 3 &4) were detected. According to these sonographic findings, the Arnold-Chiari malformation (type II) was confirmed and termination of pregnancy performed at 20 weeks of gestation. Injection syntosinone, Mifepristone and Mesoprostol were administered, spontaneous expulsion of dead fetus followed by expulsion of placenta and membranes. Dead male fetus weighing 260 grams with bilateral club foot and club hand deformity, with deformed scalp and spinal defect. Placenta weighed 110gm and liquor was clear.

The feature of the Chiari II malformation that have been most useful are the infratentorial findings, these include effacement of the cisterna magna and deformation of the cerebellum (banana sign),skull deformities (the "lemon sign"), (Fig 5). No evidence of ventriculomegaly in this case.Ventriculomegaly is considerably less common before 24 weeks than after 24 weeks in fetuses affected with myelomeningocele (8). However many case of Arnold chiari malformation have been reported, association of club hand deformity not described as far as the research we have done in the literature.

DISCUSSION

In 1891, Chiari[18] described the anomalies of the hindbrain that he found in cases of congenital hydrocephalus and divided these malformations into four categories. Arnold-Chiari malformation is a congenital malformation of the spine and posterior fossa characterized by myelomeningocele (lumbosacral spina bifida aperta) and a small posterior fossa with descent of the brainstem. Numerous associated abnormalities are also frequently encountered. Multiple studies have evaluated the accuracy of sonography for diagnosis of Chiari malformation (5-7).

The feature of the Chiari II malformation that have been most useful are the infratentorial findings, these include effacement of the cisterna magna (5) and deformation of the cerebellum (banana sign), although other infratentorial abnormalities are commonly observed postnatally (6-7). Many supratentorial abnormalities have also been described in literature in arnoldchiarimalformation(6-7). These supra tentorial abnormalities are callosal dysgenesis, a small third ventricle, a beaked tectum ,enlarged interthalamic adhesions, heterotopias, polymicrogyria, , skull deformities (the "lemon sign"), colpocephaly, and other cause of ventriculomegaly. One of the Importantfindings is ventriculomegaly because visualization of the lateral ventricle is required on all routine sonography. (8).inour case, lemon shaped skull was identified. Lateral ventricles were within normal limit. Ventriculomegaly was not demonstrated in this case, may be due to imaged earlier. ventriculomegaly is considerably less common before 24 weeks than after 24 weeks in fetuses affected with myelomeningocele. In our study, no supra ventricular anomaliesidentified, other than lemon shaped skull.

The severity of posterior fossa (PF) deformity was graded to mild, moderate and severe. The PF deformity was considered mild when smaller than normal (<2mm) but identiable. Cisterna magna was present and cerebellum was large enough to identify and appear not misshapen.A moderate deformity was diagnosed when the PF subjectively appeared somewhat small, the cisterna magna was effaced and misshapen cerebellar tissue could confidently be identified (banana shape) (Fig 5). The PF deformity was considered severe when PF to be very small, the cisterna magna was affected and little or no identifiable cerebellar tissue was visible (8). According to this grading system, our casehas moderate PF deformity.

The cranial findings associated with the Chiari II malformation are found exclusively in fetuses with myelomeningocele. Therefore, identification of the Chiari II malformation virtually ensures that myelomeningocele is present. Probably, among these supratentorial findings are the so called lemon sign (inward scalloping of the frontal bones) and ventriculomegaly. However, the lemon sign is frequently not present in later pregnancies (9) and can be seen in healthy fetuses (9, 10) and in other conditions (10). In addition, ventriculomegaly may be absent particularly before 24 weeks (8) but when is present, is nonspecific for myelomeningocele (11).

Other skeletal anomalies described in literature were as mentioned below:

Spinal anomalies weresyringohydromyelia, scoliosis in adult patient, segmentation anomalies (klippelFeil syndrome, atlanto axial assimilation)diastematomeylia , scalloping of petrous , temporal bone, enlarged foramen magnum, Luckenschadel skull, small posterior fossa. Associated limb anomalies are club foot.In our case, we could demonstrate lemon shaped skull , bilateral club foot and bilateral club hand.Among the associated limb anomalies, club hand have not been mentioned in the literature as far as our knowledge concerned.The diagnosis of myelomeningocele in a fetus is important , since It provides the parents with an opportunity to consider pregnancy termination .Among parents electing to continue the pregnancy, adequate counseling and psychological preparation can be provided (15). In our patient, more accurate evaluation on spinal canal was performed after detection of "Lemon sign" and posterior fossa anomaly at 19 weeks of pregnancy. In conclusion, the ultrasonography prenatal screening is emphasized as the primary method of assessment of the early fetal malformation. Early diagnosis of such malformation helps to make decision to offer further fetal karyotyping or termination of pregnancy.

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