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Prenatal diagnosis of Arnold-Chiari syndrome using multi-slice view 3D/4D ultrasound and MRI

Robert Dankovcik[1], Eduard Kucera[2], Lydia Frigova[3], Patrik Stovcik[4], Anna Plichtova[5], Marek Dudas[5]

[1]2nd Department of Obstetrics and Gynecology, P. J. Safarik University and L. Pasteur University Hospital, Kosice, Slovak Rep., [2]Mother and Child Care Institute Praha-Podoli, Czech Rep., [3]Department of Radiology, Safarik University, Kosice, Slovak Rep., [4]Clinic of Pediatric Neurology, Presov, Slovak Rep., [5]Fetal Medicine Program, Faculty of Sciences, P. J. Safarik University in Kosice, Slovak Rep.

Correspondence: Robert Dankovcik, MD, PhD, 2nd Department of Obstetrics and Gynecology, L. Pasteur University Hospital, Rastislavova 43, SK 04001 Kosice, Slovak Republic, E.U., Tel.: +421 905 359 381, e-mail: dankovcik@mail.pvt.sk

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Case report

Abstract
Arnold-Chiari malformation is a congenital deformity of rear brain, i.e. anomaly of the lower part of cerebral stem and cerebellum involving their caudal displacement through foramen magnum into the spinal canal. Early diagnosis and adequate treatment may be decisive for further psychomotor development of an affected individual. Here we present a case of a state-of-the-art prenatal diagnosis of Arnold-Chiari syndrome using 3D extended imaging, 3D/4D ultrasound and fetal MRI-like imaging, together with a literature review of this anomaly rarely diagnosed in prenatal period.

Key words:
Arnold-Chiari malformation, magnetic resonance imaging, prenatal ultrasonography

PRENATÁLNÍ DIAGNOSTIKA ARNOLD-CHIARIHO SYNDROMU POUŽITÍM MULTI-SLICE VIEW 3D/4D ULTRAZVUKOVÉHO ZOBRAZENÍ A MRI

Kazuistika

Abstrakt
Arnold-Chiariho malformace je vzácná kongenitální vada zadního mozku, tj. anomálie dolní části mozkového kmene a cerebella, které se posunují přes foramen magnum kaudálně do páteřního kanálu. Časná diagnostika a adekvátní léčba jsou důležité pro další psychomotorický vývoj postiženého jedince. Prezentujeme případ prenatální diagnostiky Arnold-Chiariho malformace použitím 3D/4D uzltrazvukového zobrazení a fetální MRI-simulovaného zobrazení včetně přehledu této vady, jen vyjímečně diagnostikované v prenatálním období.

Klíčová slova:
Arnold-Chiariho malformace, magnetická rezonance, prenatální ultrazvuková diagnostika

Introduction

The Arnold-Chiari malformation may include multiple forms that may escape clinician's attention during prenatal diagnosis. This article provides an example of state-of-the art imaging of this anomaly in the fetal period. For the first time in the history this malformation was described by Cleland in 1883, followed by Chiari in 1891 and subsequently in 1894 by Arnold. These authors were first to emphasize the importance and to summarize at least partially the most prominent features of this anomaly (1). Chiari describes 4 basic types of the malformation:

- type I – herniation of cerebellar tonsils,
- type II – herniation of cerebellum and lower part of the brain stem,
- type III – rare finding of herniation of the brain stem together with cervical or occipital encephalocele,
- type IV – extreme cerebellar hypoplasia with caudal displacement of the contents of rear brain.

Case Report

36-year old woman in third pregnancy and a history of two deliveries was referred for ultrasound assessment in 32nd week of pregnancy based on a suspicion of dilated lateral ventricles **(Figure 1)**. Before referral, the patient was examined by ultrasound in 15th and in 22nd week of pregnancy with negative morphological findings, negative biochemical screening, AFP 1.62 MoM. There was nothing of significance in the family and personal history. Previous two pregnancies were uncomplicated including labor and puerperal period. Ultrasound examination yielded the following findings: femoral measurement and fetal abdominal circumference corresponding to gestation age (32nd week). Biparietal diameter (BPD) and fetal head circumference correspond to 27th week of pregnancy. Typical „lemon sign" fin-

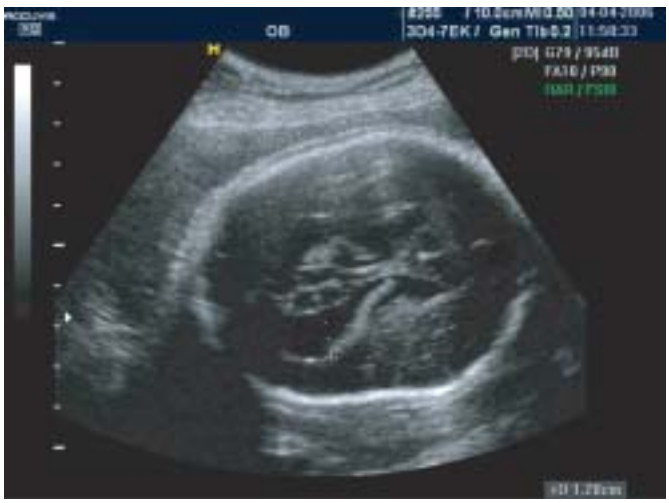

Figure 1 *Mild dilatation of the lateral ventricule.*

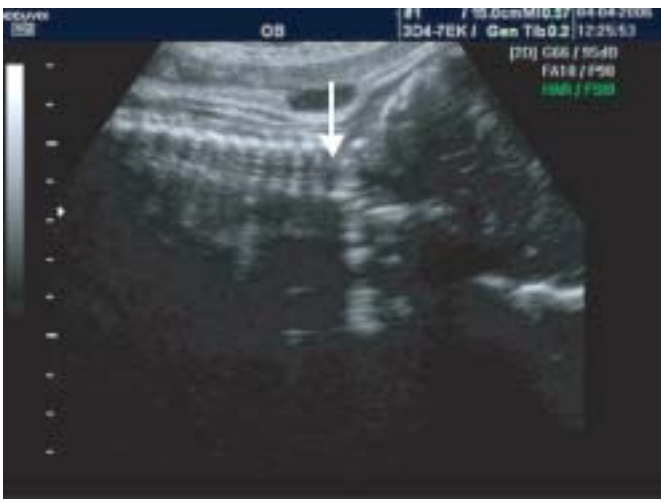

Figure 2 *Herniating posterior fossa contents into upper cervical canal.*

Table 1 *Classification criteria of Chiari malformation type II (2,3,4,5).*

Anatomic locality	Accompanying anomaly
Posterior cranial fossa	Hypoplastic with widened foramen magnum, ponto-cerebellar angle and cisterna magna (cerebellomedullary cistern) are obliterated, cerebellar tentorium is positioned low and at acute angle
Cerebellum	Herniation of cerebellar tonsils and vermis cerebelli – displaced distally through foramen magnum into cervical spinal canal and also upward herniation through tentorial incisure (so called "towering cerebellum"), embracing the pons from the rear (so called "heart or banana sign") including herniation into cerebellopontin angles
Brain stem	Herniation of medulla oblongata and lower part of pons into cervical spinal canal, more prominent dorsally, antero-posterior narrowing of pons
Middle brain	„beak-like" deformity of tectum, aqueduct stenosis and gliosis
Ventricle system	4th ventricle small even indistinct, vertically elongated and displaced distally, 3rd ventricle dilated – of atypical bi-concave shape "hour-glass shape" with prominent interthalamic adhesion and large massa massa intermedia, frontal cornices of lateral ventricles are of „bat wing" shape, with compression of nucleus caudatus, occipital cornices of lateral ventricles are of colpocephalic shape, occlusion or atresion of the foramen Luschkae and foramen Magendie with occlusion of basal cisternas, obstruction 3-ventricle hydrocephalus (90–98 %)
Midline structures	Dysgenesis of corpus callosum (80–95 %), agenesis of septum pellucidum, hypoplastic or fenestrated falx cerebri with gyral interlocking.
Cortex	Stenogyria, polymicrogyria, cortical heterotopy, cortical heterotopy, reduction of the number of cortical, histological layers, dysplasia of gyrus cinguli
Spinal cord	Spinal dysraphism – lumbar myelomeningocele (95–98 %), low-positioned, often so called "tethering" conus medullaris, caudal displacement of cerfical spine with so-called. Cervicomedullar „kinking" with traction of denticulate ligament in the area of C2-C4 (70 %), cervico-thoracic syringohydromyelia, diastematomyelia, cranial transposition and compression of upper cervical spine nerve roots
Skull	„Luckenschadel" skull (85 %)-prominent frontoparietal part of the skull due to dysplastic membranous ossification which disappears after 6 months of age, cranio-facial deformation and face asymmetry, bone defects of skull – so called „foramina" bifrontal, craniolacunar pattern on the surface of clivus and petrous resulting from pressure of cerebellum, dilated subarachnoidal space in frontal part frequently with fibrous adhesions, low positioned hairline

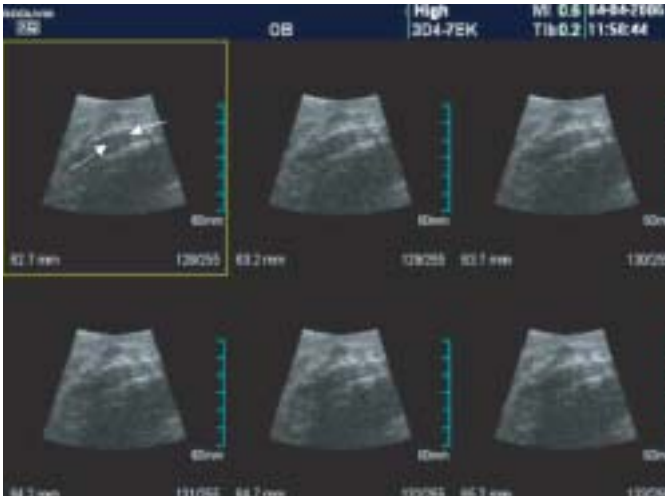

Figure 3 *Multi – slice view of the meningomyelocele.*

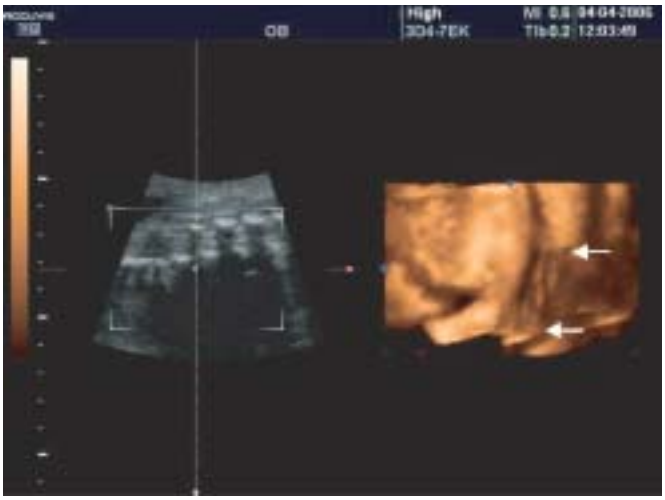

Figure 4 *3D of the meningomyelocele.*

ding. Width of lateral ventricles at atrial level is 1,45 cm with typical „dangling" choroid plexus. 3rd ventricle is dilated and elongated across front-to-rear axis. Atypical finding in posterior cranial fossa , whereas it is impossible to identify the cerebellum. Tentorium arms are close to each other, cerebellum "escaping" distally. As it is impossible to acquire image of cerebellum by conventional 2D imaging, the area of cervical spine and posterior cranial fossa is examined using 3D/4D ultrasound (ACCUVIX XQ Medison). Volume data have been transformed into sequence of 2D slices (Multi – Slice ViewTM), which enable viewing of the scanned area in multiple slices similar to sonographic MRI. A series of slices has confirmed protrusion of cerebellar hemispheres below the level of C2 **(Figure 2)**. Continued examination towards distal parts has discovered lumbo-sacral meningomyelocele **(Figure 3)**, confirmed in 3D/4D mode **(Figure 4)**. Arnold-Chiari syndrome type II was confirmed by ultrasound. Subsequent MRI scan (Siemens Symphony Maestro Class) **(Figure 5)** has confirmed the above diagnosis. The patient delivered in 38th week by C-section. Female fetus weighing 3110 grams /48 cm, Apgar 10 in 1 min, 10 in 10.min. On the day of delivery the meningomyelocele was resected and reconstruction was carried out. Post-operative course was without complications, the mother and the child

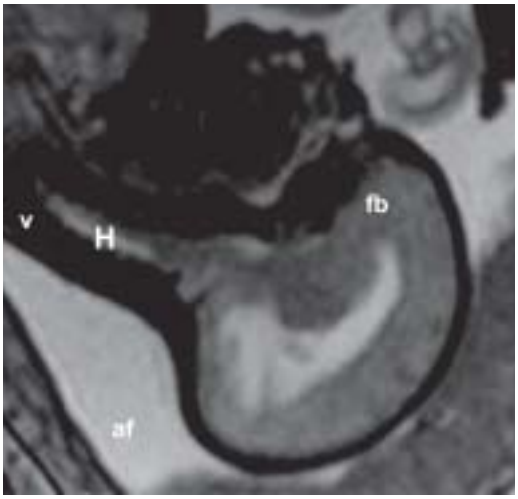

Figure 5 *MRI of the fetus – the posterior fossa contents is herniating into the upper cervical canal. H – Herniation; v – vertebral column; fb – forebrain; af – amnionic fluid*

were dismissed on 6th day post partum.

Discussion

In 1894 Julius Arnold described a child with spina bifida and meningomyelocele with a theory of its development. Arnold only assumed that abnormalities of cranial parts of CNS are likely to accompany this kind of spinal dysraphism . In that case, hydrocephalus was not observed, even though in 1896 Chiari likened this case to his type II with hydrocephalus which he suspected also in Arnold?s case and therefore they jointly termed this 2nd type as Arnold-Chiari malformation. All other malformations are simply Chiari malformation even though many literary sources and clinical medicine does not strictly adhere to this terminology and use of both authors? names is preferred for all four types of this complex anomaly – see Table 1 for classification criteria.

Genetic diagnosis of craniofacial anomalies based on prenatal or preimplantation testing /6/ is peculiar due to an insufficient palette of genetic markers, while emerging technologies such as experimental high-resolution MRI (7) bring new perspectives into visual detection of malformations. Since 1980, the MRI in sagittal projection and T1-weighting, in addition to precise history and detailed neurologic examination, has become the method of choice and to-date unsurpassed diagnostic standard within the imaging algorithm of Chiari malformation (8). In infants and children with open fontanel, the ultrasound is the method of first choice due to accessibility, safety and technical simplicity, even though the resolution, especially with respect to posterior fossa is not nearly at the level of MRI. Ultrasound examination of brain in this age group has become so wide-spread, that in some countries (including Slovakia) it has become a part of screening.

One of the advantages of ultrasound techniques is that it helps in diagnosis of this malformation, especially with respect to type II and III associated with myelo- and encephalocele, in prenatal phase screening. However this technique is burdened by certain bias on the part of the examiner, therefore diagnosis of these and other

anomalies in the area of CNS requires responsible approach, high level of qualification, erudition and most of all experience. Today's high-end ultrasound devices – in addition to high-resolution capability – offer new imaging options for imaging of skeletal and soft-tissue features. State-of-the-art 3D/4D techniques enable processing of a single volume 3D scan into a sequence of up to 24 2D slices. The resulting ultrasound image is similar to MRI, thus erasing borders between diagnostic ultrasound and radiological examination.

Conclusion

Children with myelomeningocele and symptomatic Chiari II malformation are still burdened by a relatively high morbidity and mortality (9,10), exceeding 50 % according to several sources, depending on treatment strategy and post-operative care/tracheostomy, gastrostomy a nasogastric probe are essential for several months since the procedure/. Early diagnosis of this congenital malformation is important for adequate management of pregnancy and scheduling of C-section delivery at an institution close to neurosurgical clinic.

Acknowledgement

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