

Report of a case of spinal dysraphism with lemon and banana signs in the second trimester and normal intracranial translucency in the first trimester

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1. Introduction:

All cases of open spinal dysraphism (OSD) are associated with Arnold-Chiari malformation as a consequence of leakage of cerebrospinal fluid into the amniotic cavity leading to caudal displacement of the hindbrain. Typical cranial signs (lemon and banana signs, ventriculomegaly, microcephaly) are then detected during second-trimester ultrasound scan. Conversely, they are absent in all types of closed spinal dysraphism (CSD) (1). In 2009 Chaoui et al. (2) reported a new first-trimester marker called intracranial translucency (IT), easily detectable in the same mid-sagittal plane of the fetal face as for the measurement of nuchal translucency (NT). In normal fetuses IT corresponds to the fourth cerebral ventricle, while in fetuses with OSD it is suggested there may be absence of the IT due to caudal displacement of the brain resulting in compression of the fourth ventricle. We report a case of fetus with normal IT in the first trimester and at the same time with the development of Arnold-Chiari malformation later in the second trimester.

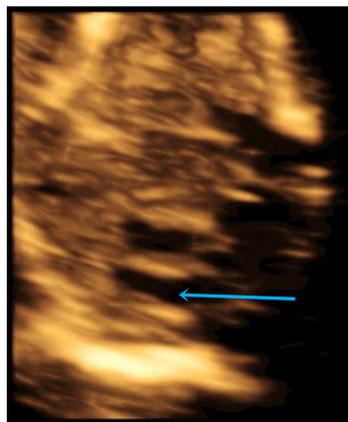
2. Case report:

A 31 year old primigravida presented at our clinic for the first trimester screening in 13 weeks. The NT was increased to 5,1mm (3,18MoM) and there was a small omphalocele, micrognathia and fetal edema. The risk for trisomy 21 was 1:20 and for trisomy 18/13 1:7. CVS was performed with normal karyotype (46,XY). In 16 weeks we repeated ultrasound scan with the findings of Arnold-Chiari malformation (lemon sign, banana sign, atrial width 8mm), omphalocele, contractures of upper extremities and clenched hands. We were not reliably able to localize the corresponding spinal defect. Because of a very strong suspicion to Edwards syndrome, we opted for iterative invasive testing. Amniocentesis was performed with normal karyotype (46,XY) and the level of amniotic-fluid AFP of 16,4 IU/l (1,72MoM). After counseling partners elected for termination of pregnancy. Anomalies noted at autopsy included CSD in the sacral region (skin-covered spinal cord), omphalocele, contractures of extremities with fibrous bands, ventricular septal defect, low set ears, receding chin. We retrospectively reviewed stored images and 3D volumes from the first trimester scan and found normal intracranial translucency and normal fourth ventricle.

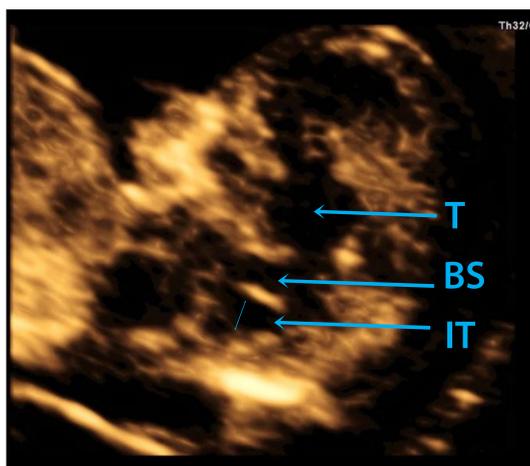
First trimester ultrasound images (13. weeks of gestation)



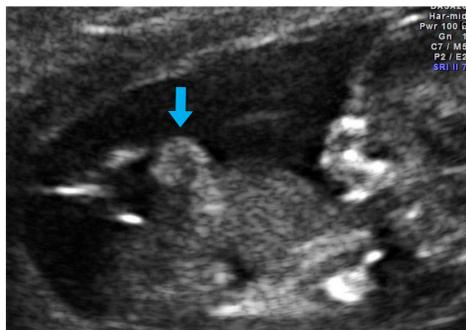
Increased NT and micrognathia



Transverse view at the level of fourth ventricle (3D VCI)

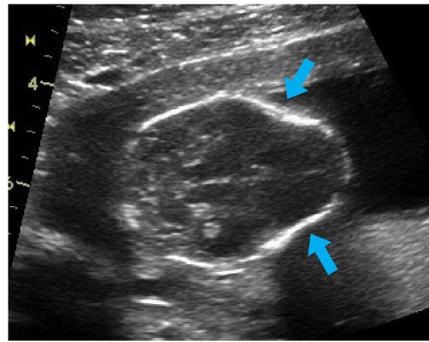


Mid-sagittal view of fetal face with thalamus (T), brain stem (BS) and normal intracranial translucency (IT) (3D VCI)

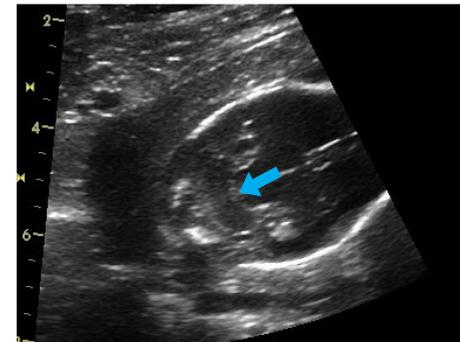


Small omphalocele

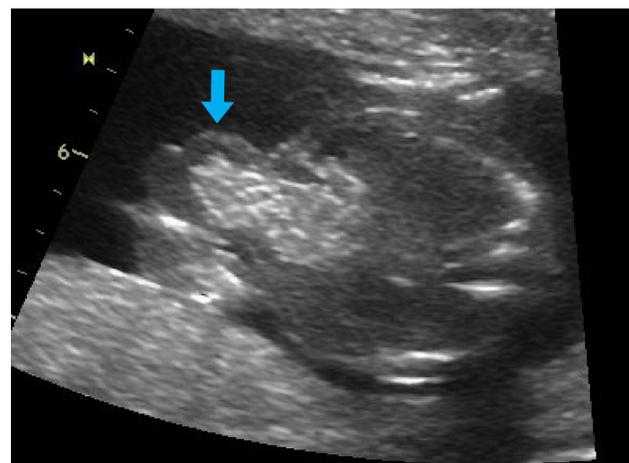
Second trimester ultrasound images (16. weeks of gestation)



Lemon sign



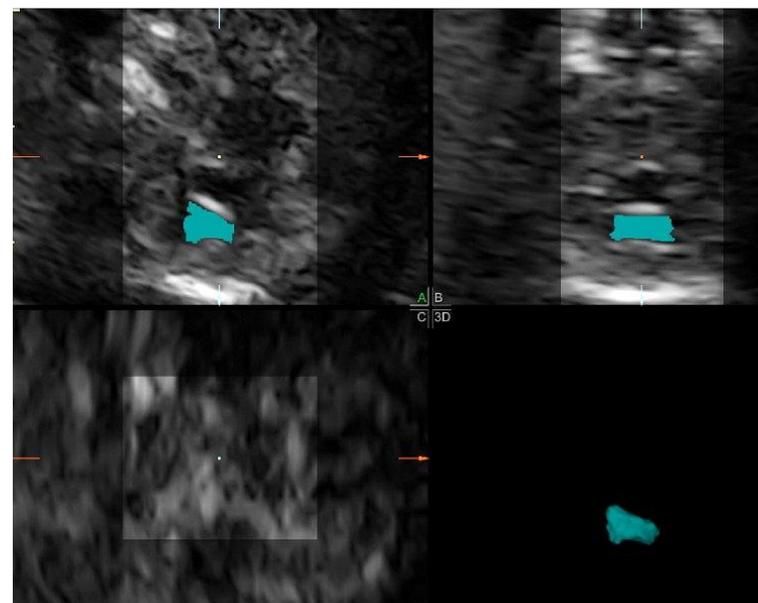
Banana sign



Omphalocele containing bowels

3. Discussion:

We observed seemingly inconsistent combination of second-trimester signs of Arnold-Chiari malformation and normal first-trimester IT. The possible explanation of normal IT is that the leakage in the first trimester is probably small or not sufficient enough for caudal brain displacement and compressing the fourth ventricle. Later it increases resulting in Arnold-Chiari malformation; however, the normal AF-AFP indicates cerebrospinal fluid leaks not into amniotic cavity. The possible mechanism of cerebrospinal fluid leakage in CSD explains exceedingly rare condition of dorsal enteric fistula (1), consisting of the cleft connecting the bowel with the dorsal skin surface through the prevertebral soft tissues, vertebral bodies, spinal canal, neural arch and subcutaneous tissues. Dorsal enteric fistula is due to failure of notochordal integration with full-thickness persistence of the neurenteric canal. There is reportedly a strong association with other malformation of viscera (e.g. OEIS complex) (3). This condition most probably corresponds to our case.



3D reconstruction of the fourth ventricle (3D AVC)

4. Conclusion:

We observed a case of fetus with developed Arnold-Chiari malformation in the second trimester and normal intracranial translucency and fourth ventricle in the first trimester.

Reference :

1. Tortori-Donati P, Rossi A, Cama A. Spinal dysraphism: a review of neuroradiological features with embryological correlation and proposal for a new classification. *Neuroradiology* 2000; 42: 471-491.
2. Chaoui R, Benoit B, Mitkowska-Wozniak H, Heling KS. Assessment of intracranial translucency in the detection of spina bifida at the 11-13-week scan. *Sultrasound Obstet Gynecol* 2009; 34: 249-252.
3. Hoffman CH, Hankins L, Kramer L, Wilson BA. The split notochord syndrome with dorsal enteric fistula. *Am J Neuroradiol* 1993; 14: 622-627.

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