Pictorial essay

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Prenatal ultrasound diagnosis of neural tube defects. Pictorial essay

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Abstract

Neural tube defects (NTD) are a heterogeneous group of malformations resulting from failure of normal neural tube closure before the fourth and fifth week of embryologic development. The three most common forms of NTD are: anencephaly, encephalocele and spinal dysraphism. Less common forms of neural tube defects include iniencephaly, amniotic bands and other types of spinal abnormalities including scoliosis/cyphosis, sacral agenesis, limb-body wall complex, diastematomyelia. The most part of these abnormalities are accessible to the ultrasound diagnosis in the midtrimester and sometimes even in the late first trimester of the pregnancy. This kind of abnormalities can occur in isolation or in association with other anomalies, which can also be characterized with ultrasound. In this pictorial essay the ultrasonographic aspects of the NTD will be discussed.

Keywords: neural tube defects, anencephaly, encephalocele, meningomyelocele, spinal dysraphism.

Introduction

The inicidence of neural tube defects (NTDs) is approximately 1 to 2 per 1,000 birth. Anencephaly and spinal dysraphism are most common with nearly equal prevalence of 1 per 1,000 [1].

Acrania/Anencephaly. The incidence is approximately 1 per 1,000 birth. Spinal dysraphisms, cleft lip and palate, urinary tract, gastrointestinal and cardiac abnormalities may be associated. Polyhydramnios is common after 25 weeks. At ultrasound (US), the most striking feature of acrania in the second and third trimester, best appreciated in a coronal image of the face, is absence of the calvaria above the bony orbits that appear prominent. The ventricles and thalami are not visualized. With US there are detection rates of 100% of acrania during the midtrimester; detection of acrania is also possible early in gestation, by 11 weeks. Vaginal sonography may be necessary when the fetal head is low in the pelvis. This may be considered as two subtypes: exencephaly and anencephaly. With anencephaly, the absence of the cerebral hemispheres is found, little to no cerebral tissue is recongnized, although small amounts of tissue may be seen protruding from the defect. Exencephaly shows absence of the calvaria cephalad to the orbits but with relatively normal amount of abnormally developed cerebral tissue. Exencephaly observed during the first trimester tipically evolves into anencephaly [1] (fig 1).

Amniotic Band Syndrome. The syndrome is a destructive fetal complex caused by disruption of the amnion. At US, amniotic band syndrome should be considered whenever unusual types of clefts (involving more than one area), asymmetrical spinal defects, cephaloceles, or amputation defects of extremities are identified; intra-abdominal wall defects, asymmetrical constriction of limbs or digits, club feet, scoliosis or thoracic deformities may be associated [1] (fig 2).

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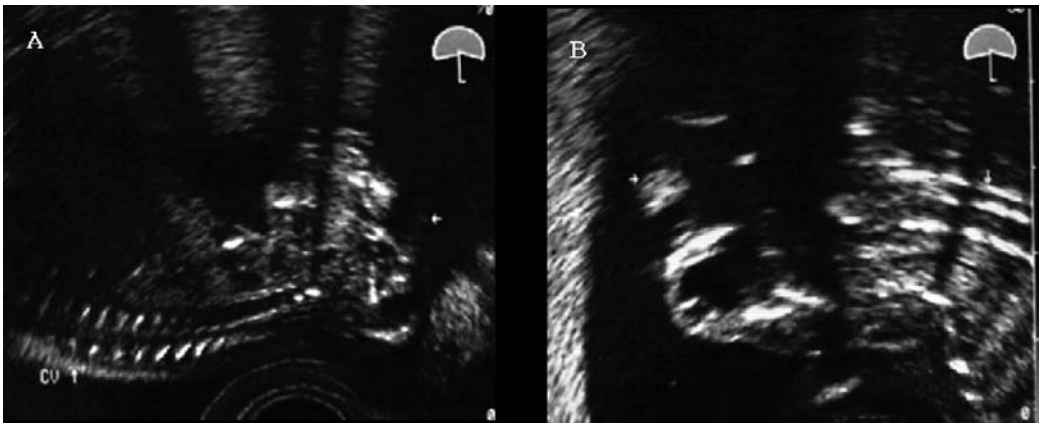

Fig 1. Second trimester fetus (19 weeks) with anencephaly. A: Sagittal and B: Coronal transvaginal scans show anencephaly with absence of brain and calvaria cephalad to the orbits.

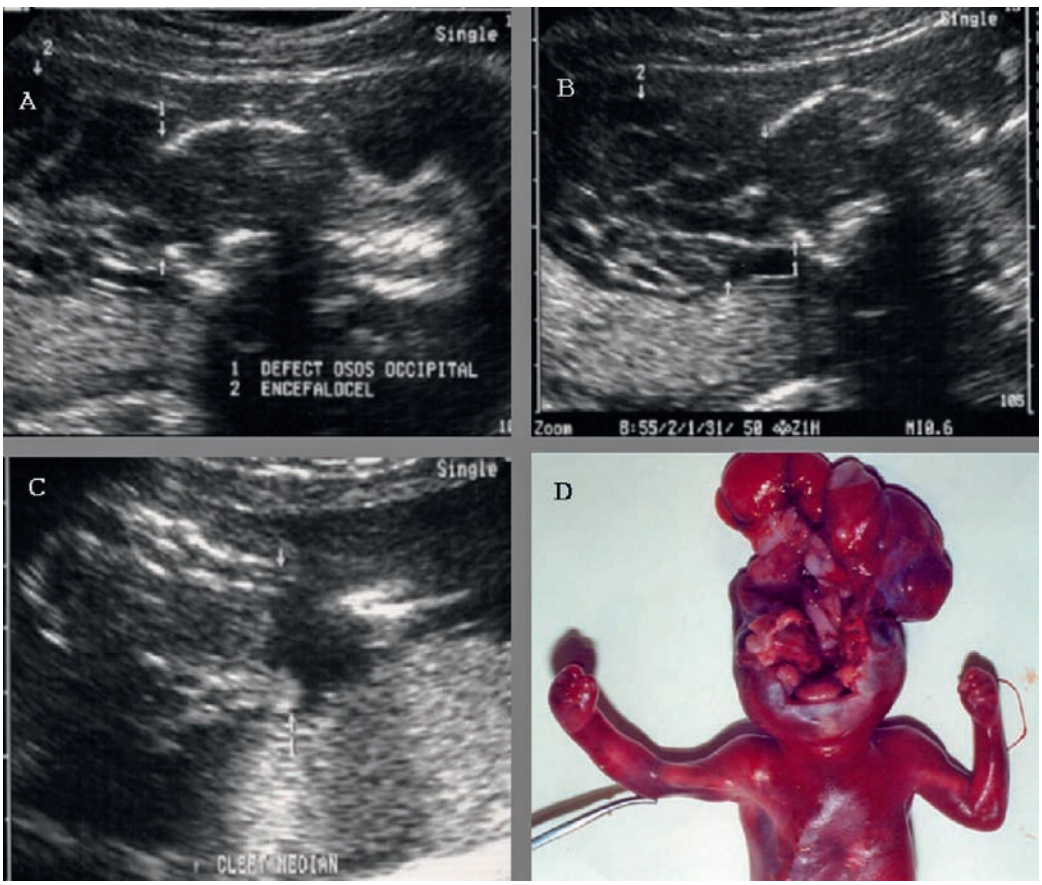

Fig 2. Amniotic band syndrome. A: Coronal scan shows large cranial defect (1); large amount of exposed brain (2), protruding through the defect is identified. B: Axial scan shows median facial cleft (arrows) showing that this is not typical anencephaly alone. C: Pathologic photograph of the case shows protruding brain through the cranial defect and facial cleft secondary to amniotic bands; the eyes are also involved and separated laterally; amniotic band is seen adherent to the fetal hand.

Cephalocele. Cepahaloceles are protrusions of intracranial structures through a defect in the skull. The incidence varies between 1 to 3 per 10,000 to 1 in 5000 live birth. According to the site of the lesion, cephaloceles can be classified as occipital, parietal, and anterior (with frontal and basal varieties). According to the content of the lesion, cephaloceles are classified as meningocele (contains meninges only), encephalocele (contains brain tissue only), encephalomeningocele (contains meninges and brain tissue) and encephalomeningocystocele (contains meninges, brain tissue and lateral ventricles). Ventriculomegaly, spinal dysraphism, microcephaly, chromosomal aberrations may be associated. Frontal cephaloceles are often associated with median cleft face syndrome, characterized by hypertelorism and median cleft lip or palate. Most cases are easily diagnosed on cranial views with brain protruding through a midline occipital defect. Other cephaloceles are small and could be overlooked. Some cases of cephaloceles have been detected as early as the first trimester, especially those associated with Meckel-Gruber syndrome. The US appearance of the lesion may change throughout gestation. Transition from a solid to a fluid pattern and transient disappearance heve been described. Ventriculomegaly, frontal bossing, obliteration of the cisterna magna and distortion of normal cerebral landmarks may be associated. Displacement of the cerebellum inside the cepahlocel is occasionally observed, and is referred to as Chiari`s deformity type III. Hypertelorism and distortion of intracranial morphology may suggest the diagnosis of anterior cephaloceles [1] (fig 3).

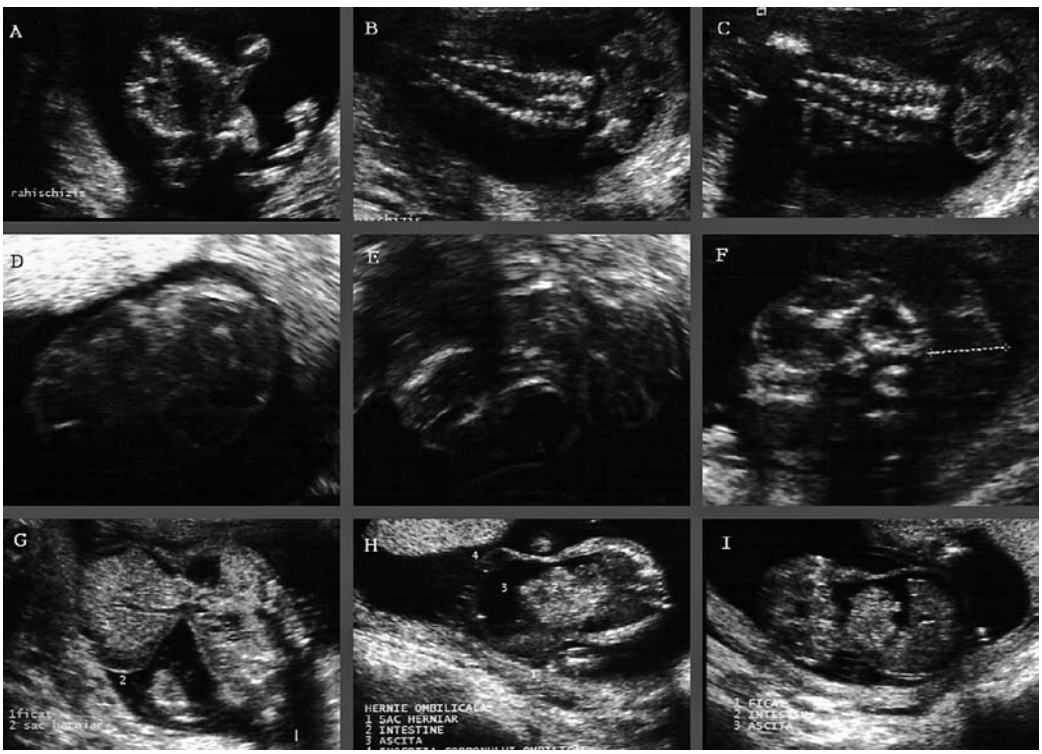

Fig 3. Second trimester fetus (18 weeks and 5 days) with large encephalocele, cervicothoracal rachischizis and liver-containing omphalocele. A: Axial ultrasound in the thoracic spine showing a full thickness defect of the soft tissue overlying the spine and flat nonprotruding defect. B, C: Coronal views demonstrating scoliosis and lateral splaying of the lateral processes with widening of the spinal canal of the cervicothoracic spine (cervicothoracal rachischizis). D,E: Transvaginal scans of cranium show large amount of brain protruding through the large cranial defect. F: A transverse scan in the posterior cervical region demonstrating increased nuchal fold: 12,8mm (normal range in second trimester normal fetus 6mm). G, H, I: Omphalocele with extracorporeal liver. G: A transverse scan of the abdomen shows an omphalocele sac containing liver (extracorporeal liver); the omphalocele sac is central and well contained by the surrounding membrane; the omphalocele sac is larger than the central defect. (2) – omphalocele sac; (1) – extracorporeal liver. H, I: Coronal scans showing prolapse of small bowel loops into the omphalocele sac and ascites. 1 – omphalocele sac; 2 – bowel loops; 3 – ascites; 4 – insertion of the ombilical cord in the centrum of the omphalocele sac.

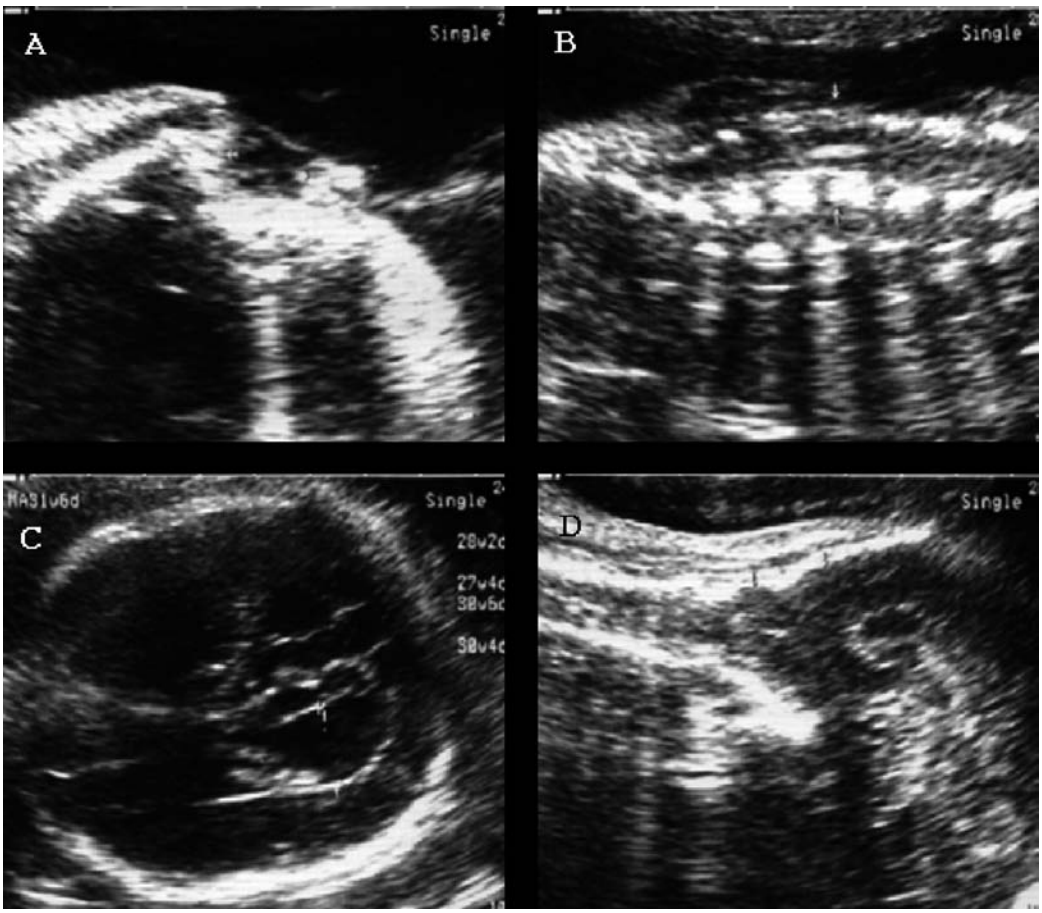

Fig 4. Open spinal dysraphism during the third trimester (31 weeks and 5 days). A: Axial ultrasound through the spine demonstrating a full thickness defect of the soft tissue overlying the spine; U shaped vertebra with lateral splaying of the lateral processes with cystic appearance of the meningomyelocele. B: Coronal scan through the spine demonstrating lateral splaying of the lateral processes with widening of the spinal canal (arrows). C: Axial ultrasound in the fetal head demonstrates ventricular moderate bilateral dilatation (14mm). D: Sagittal ultrasound in the posterior fossa shows Arnold-Chiari malformation with inferior herniation of cerebellar vermis.

Spinal dysraphisms. The incidence is approximately 1 per 1,000 [1]. The defect is located in the lumobsacral region in 90% of cases, in the thoracic region in 6-8%, and in the cervical vertebrae in 2-4% [2]. There is a defect of the vertebrae that may be posterior, anterior (rarely) or, in rare cases, consists of a splitting of the vertebral body [1]. Spinal dysraphisms are subdivided into open spinal dysraphisms (OSDs) and closed spinal dysraphisms (CSDs). CSD (15% of cases) consists in a small defect completely covered by skin [3]. With OSDs (85% of the dorsal defects) the vertebrae are lacking the dorsal arches and the nervous tissue is exposed to the environment through a congenital skin defect [1]. According to the content of the lesion spinal dysraphisms are classified as meningocele if it contains meninges only or as meningomyelocele if it contains meninges and neural tissue. With myeloschisis the spinal cord is widely opened dorsally and is part of the wall of the meningomyelocele. Rarely, a meningocele may be covered by intact skin. Arnold-Chiari malformation and foot deformities (like dislocation of the hip, clubfoot, rockerbottom foot) can be associated [1]. To empahsize the defect, sagittal, coronal and axial ecographic scans are used. In the sagittal plane, with OSDs, the posterior line (formed by the ossification centers of the posterior elements of the vertebrae) and the overlying soft tissues are absent at the level of the lesions [4]. Sagittal scans can also be used to determine the level and the extent of the lesion [5]. In the coronal plane, with OSD the central line (corresponding to the ossification centers of vertebral bodies) disappears and the two external lines

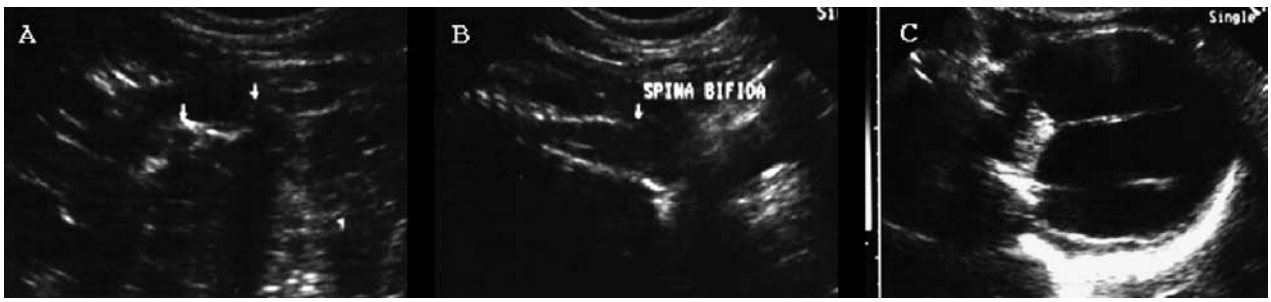

Fig 5. Open spinal dysraphism in third trimester (37 weeks). A: Axial ultrasound in the fetal spine demonstrating the full thickness defect of the soft tissue overlying the spine and the U-shaped vertebra with lateral splaying of the lateral processes (arrows), meningomyelocele. B: Coronal plane demonstrating lateral splaying of the lateral processes with widening of the spinal canal. C: Coronal view through the fetal head shows marked ventricular dilatation.

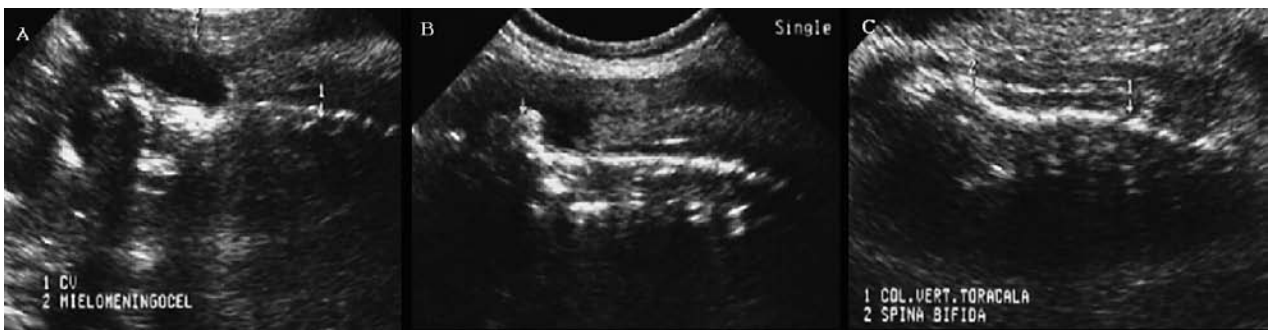

Fig 6. A 35 weeks fetus with open spinal dysraphism. A, B: Sagittal sections in the fetal spine show the cystic aspect of the defect. C: Sagittal section in the fetal spine shows the ultrasound appreciation of localization and extent of the defect.

(corresponding to the posterior elements of the vertebrae) are widened. In the transverse section, with OSD, the ossification centers corresponding to the lateral processes are splayed apart, and the neural canal is exposed posteriorly [4]. The presence of a meningomyelocele sac certainly aids the diagnosis of OSD. CSDs are extremely difficult to diagnose, with the possible exception of the rare cases associated with a large subcutaneous lipoma. [1]. In almost all cases of OSD there is associated Arnold-Chiari malformation, caused by caudal displacement of the brain and characterized by the lemon and banana signs [6]. The banana sign defines the obliteration of the cisterna magna and small size and abnormal shape of the cerebellum and the lemon sign describes the frontal bossing. A variable degree of ventricular enlargement is present in virtually all cases of OSDs at birth but in less than 70% of cases in midtrimester; when present in the midtrimester, the ventriculomegaly is borderline or mild. The sensitivity of cranial signs in identifying spina bifida exceeds 99% [4] (fig 4-6).

In the first trimester OSD can be suspected at US by two markers: the loss of the normal intracranial translucency (IT), and the decreasing of frontomaxillary facial angle. In normal fetuses the IT that corresponds to the fourth cerebral ventricle is identified on midsagittal scan as a hypoechoic structure parallel to the nuchal translucency ; in fetuses with OSD, secondary to the downward displacement of the cerebellum, there may be absence of the IT [6]. The frontomaxillary angle is measured between the upper surface of the palate and the frontal bone in a mid-sagittal view of the face. In fetuses with OSD at 11+0 to 13+6 weeks' gestation the frontomaxillary facial angle is decreased, presumably due to the impaired development of the frontal bones [7].

An anterior sacral meningocele is an extremely rare condition that consists of a herniation of the meningeal sac into the presacral retroperitoneal space through a congenital defect in the sacrum or through the widened anterior sacral foramina; the ultrasound aspect is of a pelvic cyst located posterior to the bladder [8]. Cystic

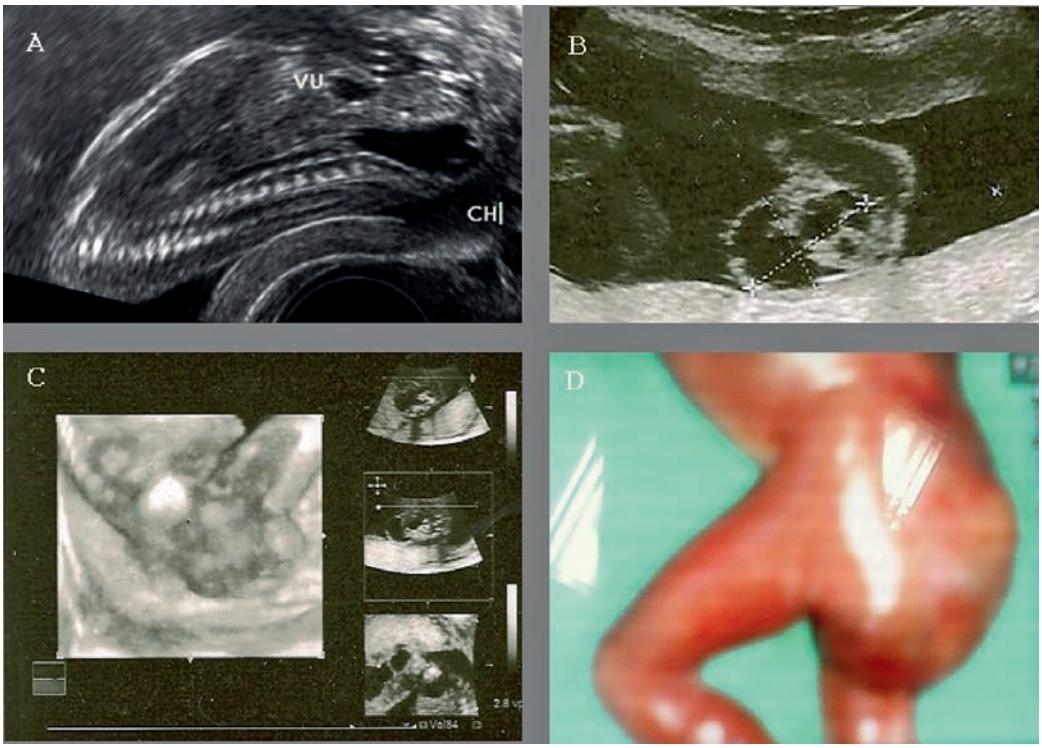

Fig 7. Growth of sacrococcygeal teratoma with cystic and solid elements. A. Parasagittal scan at 16 weeks shows cystic mass arising from sacrococcigeal area (CH). B: Follow-up scan at 21 weeks; axial scan through the sacrum shows increased growth of the mass, which has now solid and cystic elements (between calipers). C: Three dimensional ultrasound at 21 weeks shows the teratoma in three orthogonal planes plus surface-rendered view (left). D: Pathologic photograph shows the teratoma protruding inferiorly, from inside the fetal pelvis, in the right thigh.

sacrococcygeal teratomas should be considered when cystic sacral meningocele is diagnosed. The presence of cranial findings should help distinguish these possibilities in most cases [1] (fig 7).

Iniencephaly. Iniencephaly is a NTD involving the occiput and inion combined with rachischisis of the cervical and thoracic spine with retroflexion of the head with an incidence of 0.1 to 10 per 10,000 Cephalocele, holoprosencephaly, spinal dysraphism, omphalocele, gastroschisis, diaphragmatic hernia, gastrointestinal, pulmonary, cardiac, renal, skeletal anomalies may be associated. The sonographic diagnosis is made on the extreme dorsal flexion of the head, the abnormally short and deformed cervical and thoracic spine (visible on medial-sagittal scans of the spinal column), the gross alteration of intracranial anatomy, and the overall shortening of the fetus. Anencephaly or cephaloceles are present in the open forms [1] (fig 8).

With modern US equipment a large number of congenital anomalies can be consistently recognized since very early in pregnancy [9]. At 11-13 weeks it is possible to diagnose severe brain abnormalities, including holoprosencephaly, ventriculomegaly, acrania- exencephaly and encephalocele. In the first-trimester OSD can be suspected at US by an easily detectable marker within the brain: the loss of the normal IT [6]. In fetuses with OSD at 11-13 weeks' gestation the frontomaxillary facial angle is decreased and this measurement may be useful in early screening for this abnormality [7].

Conclusions

NTD are lesions that can be recognized with prenatal US in mid and third trimester but also in the first trimester, sometimes using transvaginal ultrasound. Some of these abnormalities are characterized by easily recongnizable markers that should alert the sonographer to the possibility of their presence and encourage a detailed examination for diagnosis of a specific malformation.

Conflict of interest: none

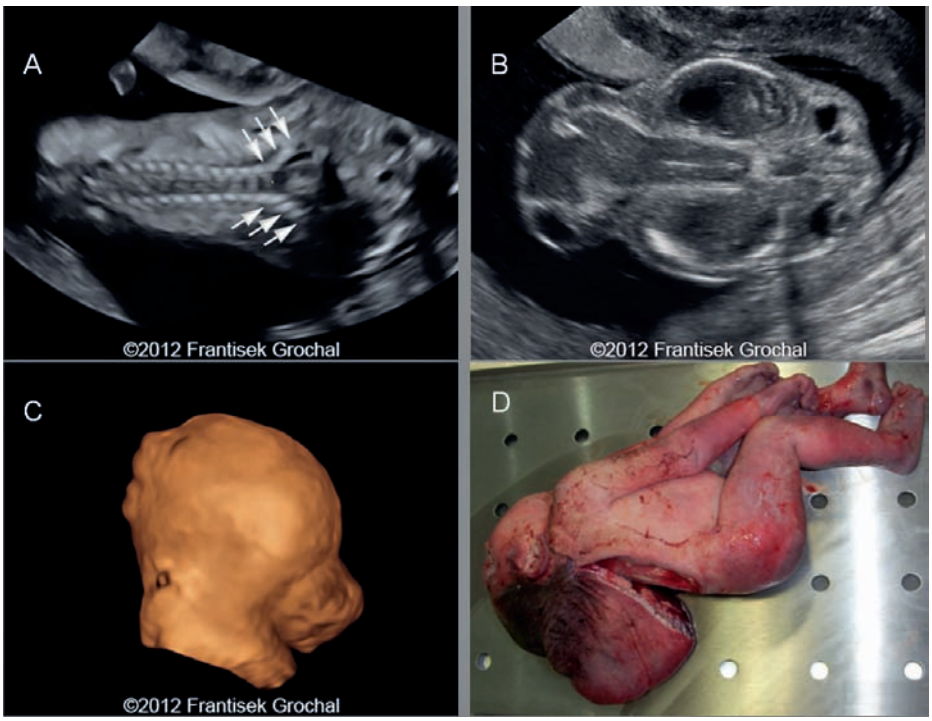

Fig 8. Iniencephaly, 14 weeks, 6 days gestational age. A. Coronal scan of the fetal spine showing large defect of the cervico- thoracic region of the spine (arrows). B. Transverse oblique scans of the fetal head showing occipital defect with a large meningomyelocele. C. 3D image showing fetal profile with meningomyelocele and permanent retroflexion of the head. D. Pathologic photograph of similar case, second trimester fetus showing occipital encephalocele and marked retroflexion of the neck. (A, B, C – Reproduced with permission from TheFetus.net)

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