

**Session 1:**

**IMAGING OF FETAL POSTERIOR FOSSA ABNORMALITIES**

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Studying the posterior fossa is challenging during the antenatal period because most of the anomalies that can be detected have a poor prognosis. A good knowledge of embryology and anatomy of the posterior fossa is mandatory to analyse the US and MR images.

**Embryological and anatomical data (1-4)**

The cerebellum is one of the first cerebral structures to differentiate but one of the last to mature, and its cellular organization continues to change after birth. Experimental studies have demonstrated a dual mesencephalic-metencephalic origin of the cerebellum whose development is closely related to that of the brain stem. Cerebellar primordia are initially lateral elements. In the dorsal region of the neural tube, the alar plate hypertrophies, forms the thickened rhombic lips over the fourth ventricle and develop towards the midline. This midline fusion in a rostral-to-caudal direction, gives rise to the vermis and is observed at the end of the 7<sup>th</sup> week. The midline fusion of the tentorium proceeds from the front to the back, so that partial tentorial defects are always posterior. During fetal life, the flocculonodular lobe is the first to be visualized, as early as the 10<sup>th</sup> week. The primary fissure separates the anterior from the posterior lobes by the 11-12<sup>th</sup> weeks so that the three main lobes (anterior, posterior and flocculonodular) are separated by the end of the first trimester. The volume of the posterior lobe is greater than the volume of the anterior lobe, with a ratio of 2:1. The posterior lobe is subdivided into three major fissures: the horizontal, the prepyramidal and the secondary fissures. The posterolateral fissure separates the posterior lobe from the flocculonodular lobe.

**Ultrasonographic analysis**

As for each anatomical region, the posterior fossa should always be imaged in the three planes of space. During the first trimester, at 8-9 weeks, one should be aware of a physiological transitory fluid-filled structure corresponding to a 2-3mm wide cisterna or rhomboid fossa located at the back of the pontine flexure (5). The cerebellar hemispheres are recognized on US from 12-13 weeks with the vermis recognizable appreciatively 1 week later. Initially, the hemispheres are hypoechoic with echogenic margins. With development of foliation, their echogenicity increases and a characteristic stripey appearance is observed by the late second trimester. The vermis is echogenic (6). The vermian fissures, notably the primary fissure, are clearly seen from 25 weeks. The posteroinferior aspect of the vermis may be open until 17.5 weeks with a majority of fetuses showing an appearance of open fourth ventricle at 13.5-14 weeks (7).

The tentorium cerebelli is not directly visible in US, but the normality of its insertion can be evaluated by assessing the position of the overlying occipital lobe (5). The cisterna magna is measured in the midline from the posterior aspect of the vermis to the internal surface of the occipital bone. It should measure between 4 and 10mm. Several fine echogenic lines may be observed within the cisterna magna, in, or near to the midline. They are thought to represent arachnoid septae (6). Sagittal views can be obtained through the posterior fontanelle with good visibility of the brain stem (8) but such an approach is not always available.

Endovaginal scanning is always contributory in cases with a deep cephalic presentation.

**MRI analysis**

On T2-W sequences, the cerebellar cortex is hypointense, the white matter is relatively hyperintense and the dentate nucleus has a low signal intensity, believed to result from a high cellularity. The primary fissure is always seen by 28-30 weeks. The visibility of the main fissures of the posterior lobe is more inconstant. Myelination is clearly detectable at the level of the tegmentum at 22 weeks. The shape of the fourth ventricle, the insertion of the tentorium cerebelli and the presence of the pontine bulge are more easily analyzed with MR than with US (2,5). The cerebellum biometry can be evaluated according to the standards published (3,5). There is proportionally a more rapid increase in vermis size than in the cerebellar hemispheres (3). It is essential to be aware of the asymptotic growth after the sixth month of pregnancy which makes it necessary to measure the transverse cerebellar diameter during the third trimester in order to detect some hypoplasias appearing late in pregnancy (9). There are no biometrical standards for the brain stem.

### **Posterior fossa anomalies (2,5,9)**

When an abnormality of the posterior fossa is detected, it is always essential to look for associated abnormalities, either cerebral or extracerebral.

#### ***Increased fluid space of the posterior fossa in US***

-With normal insertion of the tentorium cerebelli

- If there is no other abnormality (normal appearance of the fourth ventricle, normal *vermis* and hemispheres, normal brain stem and of course normal supratentorial space), the diagnosis of *mega-cisterna magna* is considered. Performance of MRI is recommended because US is less reliable to assess the normality of these structures. Mega-cisterna magna is commonly observed and is considered a normal variant. Its diagnosis is based on the elimination of other diagnoses.
- If there is mass effect on the cerebellum (*vermis* and/or hemispheres) and the cerebellum is complete, a retrocerebellar *arachnoid cyst* is suspected. When it is large, it may be associated with hydrocephalus and enlarged posterior fossa. It may also be too small to create a mass effect during the antenatal period and be overlooked even by MR because it is located in a physiologically enlarged fluid space. In those cases, it may be diagnosed only after birth. Some arachnoid cysts are lateral and compress one hemisphere or the brain stem.
- If one or both cerebellar hemispheres look too small, it may be related to a *hypoplasia* (10) or to an *ischaemic and/or haemorrhagic damage* during fetal life. The *vermis* may or may not be involved. MR is performed to search for signal abnormalities suggesting damage acquired during the antenatal period.
- If the *vermis* is small but keeps a normal shape and structure with a normal fourth ventricle (all these findings are better demonstrated by MR than by US), *vermian hypoplasia* is considered. It may be isolated with normal bulge of the pons. The hemispheres are hypoplastic or spared. This diagnosis may be established late in pregnancy. *Cerebellar hypoplasia* may also be associated with involvement of the pons, which is much more easily depicted with MRI than with US.
- If the *vermis* is absent, there is a complete *agenesis*. In partial agenesis (which is more common), a part of the *vermis* is present. In US, the diagnosis is based on midline sagittal and axial slices. The fourth ventricle looks “open” and there is an abnormal communication between the fourth ventricle and the cisterna magna. It may be very difficult to differentiate a slight partial agenesis from an incompletely rotated but complete *vermis*. Spatial resolution of US and MR is not sufficient to count the nine elements of the *vermis*.

- With abnormally high inserted tentorium cerebelli

The *Dandy Walker malformation* is defined by the association of global widening of the posterior fossa with ascent tentorium cerebelli, cystic dilatation of the fourth ventricle and partial or complete vermian agenesis. The prognosis seems better in patients with preserved vermian fissures (primary fissure and prepyramidal fissure)(11), which is better visualized with MR than with US. The normality of the pons is another good prognostic criteria in the absence of associated cerebral abnormalities.

#### ***Decreased fluid space of the posterior fossa in US***

Effacement of the cisterna magna, descent of the cerebellar hemispheres with a “banana” shape of the cerebellum and bilateral indentation of the frontal bones are highly predictive of myelomeningocele.

#### ***Decreased cerebellar biometry with normal fluid space of the posterior fossa***

It includes some forms of cerebellar (*vermis* and /or hemispheres) *hypoplasia*, some forms of vermian *agenesis* and *rhombencephalosynapsis*. Rhombencephalosynapsis is an exceptional malformation defined as fusion of the cerebellar hemispheres without vermian development. In US, the cerebellum appears in the form of a single block with a single transverse folding. The transverse cerebellar diameter is significantly reduced. The fusion of the hemispheres can also be depicted by MR (6,9).

#### ***Abnormal cerebellar echogenicity***

It may be related to an *ischaemic and/or haemorrhagic damage*, a *focal cerebellar dysplasia*, a *tumour* (very rare) or a *capillary telangiectasia*. In the latter case, the lesion is hyperechoic with normal cerebellar anatomy and without mass effect. Gradient echo T2 images show a marked hypointensity. The diagnosis is confirmed after birth on post-gadolinium MR. The lesion enhances and has characteristic irregular or brush-like borders (12).

### ***Abnormalities of the posterior fossa without involvement of the cerebellum and the brain stem***

Malformations of a dural sinus with or without displacement of the structures of the posterior fossa can be observed. Meningoceles, dermoid cysts may also involve the posterior fossa. Associated abnormalities of the cerebellum must be looked for.

### **Conclusion**

Anomalies of the posterior fossa are numerous and are often difficult to describe precisely during the antenatal period because of the thinness of the involved structures and lack of spatial resolution. MRI often plays a major role in the depiction of these anomalies, whose prognosis is often poor.

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## **SONOGRAPHIC IMAGING OF THE POSTERIOR FOSSA**

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Agensis of the cerebellar vermis is an uncommon defect, and it is usually detected while evaluating the posterior fossa. It could be as part of the DWM or as an isolated finding. It could also present as complete agensis or partial, i.e. inferior vermian agensis. It may be impossible to correctly diagnose antenatally a defect in the cerebellar vermis in all cases. A very high proportion of disagreement was found between prenatal ultrasonic diagnosis of Dandy-Walker malformation or variant and autopsy findings, emphasizing the need in accurate sonographic demonstration of the cerebellar vermis.

The purpose of our presentation is to evaluate the posterior fossa of the fetal brain.

It is emphasized, that the diagnosis of vermian agensis (especially partial agensis) cannot be made prior to 18 weeks of gestation!!

Detailed US of the posterior fossa should include not only the measurements of the vermis, but also the structure of the vermis (primary fissure, fastigium), but also the relationship to the cisterna magna, the 4<sup>th</sup> ventricle, the tentorium as well as to the pons.

Enlargement of the posterior fossa - Is it always pathological??

It depends on the gestational age. Also, the possibility of rotation of the vermis should be included in the differential diagnosis. The demonstration of the normal vermis on mid-sagittal plane in cases with "enlarged cisterna magna" on standard plane raises the possibility of rotation of the vermis.

When comparing ultrasound to MRI of the fetal posterior fossa, it is sometimes easier to perform a perfect midline sagittal slice with US than with MRI before 25 weeks. Early MRI adds little new information compared with well-conducted US.

The contribution of 3D-US to the sonographic demonstration of the posterior fossa is emphasized.

## **POSSIBLE SURGICAL SOLUTIONS FOR CYSTIC MALFORMATION OF THE POSTERIOR FOSSA**

**Dr Liana Beni-Adani, Pediatric neurosurgery, Dana Children's Hospital, Tel-Aviv, Israel**

Some of the challenging pathologies of "cystic malformations of posterior fossa" include combination of true pressure in the posterior fossa and / or associated hydrocephalus.

In the era of modern Pediatric neurosurgery, we try to minimize or at least consider options to shunting procedures of cystic pathologies.

The most important point for the surgeon is to differentiate pathologies of *malformative cystic dilatation without pressure* from *cystic malformations that cause pressure* (either in pressure in the posterior fossa or supratentorial HCP in the mechanism of blocking outlet of or compressing from outside of the 4<sup>th</sup> ventricle).

The differential diagnosis of cystic malformations of Posterior fossa have been classified by many authors in different classifications. The classification can be made on the basis of genetics, pathophysiology, outcome or practical-treatment point of view.

Choosing the last classification for invasive treatment considerations, we classify cystic pathologies in a very simple way:

1. Cystic cavity without pressure, with malformative brain.
2. Cystic malformation without pressure, with otherwise normal brain and normal cerebellum-brainstem, and (D.D. Cisterna magna).
3. Cystic malformation with pressure, and abnormal brain (malformative either to secondary insult or to primary imminent pathology of the brain – examples for each: DW, and post-cerebellar bleed).
4. Cystic malformation with pressure and otherwise normal brain (example – arachnoid cyst).

The treatment for pressure related cystic malformations of the posterior fossa depend on the clinical status of the child, the associated brain malformations, the presence or absence of hydrocephalus and the progressiveness of HCP.

In contrast to the past when all fluid pathologies associated with pressure were treated by shunts, today with modern neurosurgery anesthesia and intensive care treatment, we may consider in selected cases a heroic approach of avoiding shunts and shuntless surgeries. The options for cystic malformations in the posterior fossa remain therefore – formal craniotomy and opening of the cyst to normal CSF spaces – such as cisterns, subarachnoid space and ventricles.

For associated HCP, posterior fossa cyst treatment may resolve HCP in some cases, and for others - endoscopic 3<sup>rd</sup> ventriculostomy in addition to opening of the posterior fossa cyst may be successful.

The challenge is to differentiate between malformative not-under-pressure cavities from pressure-causing cysts, therefore the clinical judgment and not only the radiological evaluation are truly mandatory for decision taking and for proper classification. However, PRENATAL IMAGING compared to Postnatal Imaging is extremely important in understanding the pathophysiology of some cystic conditions for progressiveness of cyst and active ventriculomegaly as for the underlying possible cause of cystic formation in PF. Arachnoid cysts of the posterior fossa therefore are an excellent example for a rather "benign" cystic pathology, that will be in differential diagnosis with mega cisterna magna, and even may be mistaken on prenatal US for Dandy walker or "DW variant" (as a representative of older classification meaning cystic cavitations without tentorial elevation and with somewhat malformative components of brain in the PF). Many of those cysts will not need intervention; *however some may cause postnatal pressure on cerebellum with or without associated HCP.*

For those babies, opening of the cyst may end up with avoidance of shunt, and avoidance from shunt dependence or complications.

*We have recently learned, that* in selected patients with posterior-fossa cyst associated with supratentorial hydrocephalus, PFD may open flow-paths of CSF and enable avoiding shunts. This is true for those babies in whom the HCP was secondary to obstruction of CSF pathways in the posterior fossa.

We therefore suggest that a classification based on cystic cavity with or without pressure, and presence of malformative versus normal brain – is an important PRACTICAL classification. We also suggest that based on clinical evaluation together with radiological appearance of supratentorial ventricles and brain tissue, shuntless procedures such as isolated posterior fossa opening of cyst or with combination with ETV may be rare but important options of treatment in selected cases.

## **CORRELATIONS BETWEEN MRI AND FETOPATHOLOGY IN THE STUDY OF FETAL BRAIN STEM AND CEREBELLAR ABNORMALITIES**

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The purpose of this study was to evaluate the accuracy of MR in the description of fetal cerebellar and brain stem abnormalities. Systematic correlations between MR and fetopathological findings have never been reported in this field. A good evaluation of the abnormalities found in this area of the brain has prognostic implications and can help in parents' counselling.

25 fetuses were included in this prospective study from January 2000 to December 2005. All these fetuses presented posterior fossa abnormalities depicted by fetal MR. Termination of pregnancy was performed for all of them, followed by a thorough fetopathological examination.

MR study included evaluation of the posterior fossa and the supratentorial space, regarding biometry and morphology: visibility of primary fissure of the vermis, posterior/anterior lobe of the vermis ratio, symmetry, shape and borders of the cerebellar hemispheres, shape of the 4<sup>th</sup> ventricle, presence of the pontine budge, location of the tentorium cerebelli, evaluation of the fluid space of the posterior fossa.

Fetopathological study included evaluation of the cerebellum and brainstem weight and macroscopic and histological study of the different structures of the posterior fossa.

The MR and fetopathological criteria regarding the definition of vermian, hemispheric and brain stem hypoplasia, vermian partial or complete agenesis and destructive lesion are reminded.

A good agreement between MR and fetopathological findings was observed for the evaluation of destructive lesions, vermian agenesis and brain stem hypoplasia.

Regarding vermian hypoplasia, no false negative result was observed but there were 5 false positive results.

Regarding cerebellar hemispheres hypoplasia, the correlation was poorer since 4 false negative results and 3 false positive results could be observed.

Nevertheless, it can be concluded that, even if the structures of the posterior fossa are very thin and if there are obviously technical limitations, a thorough and systematic analysis of this region with MR can make it possible to establish most of the diagnoses.

### **Session 2**

#### **MULTIPLE CONGENITAL ANOMALIES IN PRETERM AND TERM NEONATAL POPULATION: EPIDEMIOLOGICAL STUDY WITH APPLICATION OF A TARGETED COMPUTER PROGRAM.**

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## **TITLE: FEBOTICS – A MARRIAGE OF ROBOTICS AND FETAL SURGERY**

**Michal Berris and Moshe Shoham, Technion, Haifa**

For a fetus diagnosed with a severe congenital anomaly, surgery may offer an alternative to abortion, intra-uterine death or a life with disability. Expertise is limited however, to few treatment centers worldwide and there are many technical hurdles including miniaturized instrumentation, real-time high-resolution imaging, and harmless fetal access. This article highlights various initiatives to integrate robotics into the fetal operating room. Robots assist in manipulation of delicate fetal structures by simultaneously magnifying the surgical field, filtering hand-tremor, scaling up subtle forces and introducing haptic feedback to endoscopic tools. Master-slave systems enable access to limited surgical expertise, via telesurgery. To date, three institutions have performed minimally invasive robotic fetal surgery with pregnant ewes and two human cases of instructive telesurgery have been reported. Current efforts at the Medical Robotics Laboratory at the Technion include design of a 2 mm micro-robot to insert into the pregnant uterus in order to carry out diagnostic and interventional fetal procedures. While the number of potential patients is low, research for implementation of robotics into the field of fetal surgery is justified by morbidity rates of current procedures, proven favorable outcomes with intervention, and educational value for extension to other medical disciplines.

## **CONGENITAL LUNG LESIONS (CLL): EMBRYOLOGY, PRENATAL DIAGNOSIS & MANAGEMENT**

**Laurent Garel, Professor of Radiology, University of Montreal, Canada**

The presentation aims at updating the features of CLL, especially in the recent light of antenatal recognition.

### *EMBRYOLOGY – PATHOLOGY*

- CLL result from an insult to the developing lung bud; the type of lesion is linked to the timing and severity of the causative insult.
- CLL represent a continuum of interrelated abnormalities.
- Airway obstruction appears as the common pathogenesis of CLL.
- Classification of CLL:
  1. Bronchopulmonary malformations
  2. Pulmonary hyperplasia and related conditions
  3. Congenital lobar overinflation and related conditions
  4. Systemic arterial connections to normal lung
  5. Other cystic lesions.

### *PRENATAL DIAGNOSIS*

- Spectacular increase in the number of recognized cases.
- Patterns and specificity: fetal CLL encompass a spectrum of anomalies.
- Prognostic predictors in utero: hydrops fetalis.
- Natural history of CLL:
  1. Visible on routine mid-second trimester sonogram
  2. In utero decrease in lesion size
  3. Most fetal CLL patients are asymptomatic at birth, therefore postnatal investigations are based upon prenatal findings

### *MANAGEMENT OF CLL*

- In utero: hydrops fetalis warrants fetal intervention.
- At birth: symptomatic patients need immediate surgery.
- At birth: asymptomatic patients with small and/or regressing extralobar pulmonary sequestration should be managed conservatively.
- At birth: the management of asymptomatic patients with CPAM remains controversial.
- CT is superior to chest X-ray in showing residual lesions both in the newborn and subsequently.

- Postnatal hazards of CLL have been greatly exaggerated in the surgical literature: congenital lung cysts do not degenerate to become pleuropulmonary blastomas.

### **TAKE-HOME POINTS**

- Resist the impulse to name everything.
- Select the indications for surgery.

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### **Session 3**

#### **ISCHEMIC AND HEMORRHAGIC INSULTS IN FETUSES**

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In our hospital, we have observed with fetal MR about 40 cases of haemorrhagic lesions and 50 cases of ischaemic lesions. In some cases, both types of lesions were associated.

The main physiopathological principles explaining the constitution of haemorrhagic and ischaemic damage in fetuses will be reminded.

*Haemorrhage* can involve the posterior fossa, the supratentorial and the pericerebral space. Most of haemorrhagic lesions can be detected with US and the main patterns of such lesions will be described. With MR, as for US, the appearance of haemorrhage varies according to the age of the lesion. Moreover, the type of sequence used is very important. Some old or chronic haemorrhages can be seen only with T2\* sequences. Some small parenchymal lesions can be missed by US and depicted only with MR due to its better contrast resolution. The pericerebral space is also better analyzed with MR and some haemorrhages in this space can be overlooked by US.

The type of *ischaemic lesion* depends on the stage at which the insult has appeared. In some very early damages, it may be very difficult, even with fetopathological study to differentiate an ischaemic lesion from a primitive developmental anomaly. Specificity and sensitivity of US are poor regarding the detection of ischaemic lesions. The diagnosis of ischaemia is based, with MR as with US, on both direct and indirect signs:

- the indirect signs are decrease of cerebral biometry, of active movements, polyhydramnios, in vacuo passive ventricular dilatation and Wallerian degeneration. This latter sign is much more easily depicted by MR than by US.
- the direct signs can be focal (cavitations, calcified leucomalacia, destruction of an anatomical structure) or diffuse abnormalities (abnormalities of the white matter), or cortical lesions (laminar necrosis, polymicrogyria). It must be emphasized that some ischaemic lesions (calcified leucomalacia, laminar necrosis ) can be completely overlooked by T2 sequences and showed only on T1 sequences. Therefore, it is mandatory to acquire good quality T1 images in this context. The diagnosis of diffuse abnormalities of the white matter is challenging and so far, the contribution of Diffusion Weighted Imaging has not been evaluated.

Improvement of prenatal imaging has made it possible to diagnose some haemorrhagic and ischaemic lesions which were previously overlooked and this may have judiciary implications.

## FETAL INTRA CEREBRAL HEMORRHAGE

**Haim Bassan M.D., Liana Beni Adani M.D., Shaul Harel M.D., Liat Ben Sira M.D.**  
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Fetal intracranial hemorrhage (ICH) is an acquired brain lesion with enormous impact on morbidity and mortality, as well as on long-term neurodevelopmental outcome. The diagnosis, management and outcome of fetal ICH can be expected to undergo important changes given the current advances in perinatal medicine and increased survival of premature infants. Furthermore, advanced real-time fetal US and fetal MRI techniques have enabled fetal diagnosis of ICH. Fetal ICH is increasingly recognized as more frequent as well as devastating complication in high-risk pregnancies. The exact incidence of fetal ICH is unknown. Existing neuropathological evidence suggests that fetal ICH is more prevalent than clinically appreciated at present. The exact pathogenesis of fetal ICH is unclear. As in postnatal ICH, the pathogenesis of fetal ICH is most likely related to multiple risk factors. The outcome of fetal ICH is overall poor in 40-69% of survivors, particularly in infants with severe intraventricular hemorrhage or parenchymal involvement. Most studies had a limited follow-up and did not use standardized measures to quantify global developmental performance. Therefore, in spite of the major advances in prenatal imaging of ICH, current neurological counseling, is limited by significant gap in our knowledge regarding the outcome of this important prenatal pathology.

## PRENATAL MRI IN THE DIAGNOSIS OF MALFORMATIONS OF CORTICAL DEVELOPMENT

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Cerebral cortical development (MCD) is an extremely complex process, comprising three major, but overlapping steps: cell proliferation, neuronal migration and cortical organization.

**Objective** To describe the role of prenatal MRI in the diagnosis of malformations of cortical development (MCD).

**Methods** Twenty fetuses with suspected MCD diagnosed during prenatal MR performed for various diagnoses. Their images were analyzed for their ability to define the specific malformation using an existing MCD classification by Barkovich.

**Results** The main diagnoses were: microcephaly with simplified gyral pattern, hemimegalencephaly, lissencephaly/subcortical band heterotopia spectrum, cobblestone lissencephaly, heterotopia, polymicrogyria/schizencephaly.

Associated CNS anomalies were present in all fetuses: ventriculomegaly, abnormal corpus callosum, cerebellar and brain stem anomalies, microcephaly, arachnoid cysts and abnormal skull shape. Prenatal MRI findings were confirmed by histopathological examination or postnatal MRI.

**Conclusion** Prenatal MRI had excellent correlation with histopathology findings regarding the diagnosis of MCD. We propose the use of this technique in order to confirm the diagnosis before TOP and as an alternative to histopathological examination. Prenatal diagnosed cases are usually associated with other anomalies and probably represent extreme forms of MCD.

## **NEUROPSYCHOLOGICAL OUTCOME OF CHILDREN WITH ASYMMETRIC VENTRICLES OR UNILATERAL MILD VENTRICULOMEGALY IDENTIFIED IN UTERO**

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**Objective:** To assess the neuropsychological outcome of children with asymmetric ventricles and unilateral ventriculomegaly identified in utero.

**Methods:** We assessed 21 children with asymmetric ventricles and 20 with unilateral ventriculomegaly identified in utero and compared them to a group of 20 children with symmetric ventricles, utilizing a formal neuropsychological tool: the Bayley Scale of Infant Development II (BSID-II).

**Results:** The unilateral ventriculomegaly group scored significantly lower than the control group on the mental developmental index and on the behavioral rating scale, but not on the psychomotor index. The asymmetric ventricles group did not differ significantly from the control group on either the mental or psychomotor developmental indices, but differed from the latter on the behavioral rating scale. Fifteen percent of the children of the asymmetric ventriculomegaly group performed two standard deviations below average compared to four percent of the asymmetrical ventricles group and none of the control.

**Conclusion:** Our results indicate that prenatally observed unilateral ventriculomegaly is a significant risk factor for developmental delay. The mental and motor outcome of children with asymmetric ventricles is similar to the control group but these children are at a significant risk for behavioral abnormalities.

### ***Session 4***

## **PRENATAL COUNSELING FOR CLEFT LIP & PALATE** **Leshem D., MD., Department of Plastic & Reconstructive Surgery,** **Tel-Aviv Sourasky Medical Center, ISRAEL.**

Prenatal diagnosis of cleft lip and palate can occur during routine screening obstetrical ultrasound, and high resolution ultrasound or fetal MRI done for other reasons. The affected family should be referred for consultation to a cleft lip and palate team.

The prenatal consultation includes a review of cleft lip and palate severity related to the sonographic findings. Explanation regarding cleft treatment along the years till adulthood. Viewing of preoperative and postoperative photographs of the different severity clefts. The family receives a pamphlet that gives an overview of cleft lip & palate. Any open questions can be addressed to the team members who are available for an additional meeting.

The goal of the counseling is to provide the families with the sufficient relevant information, in order to prepare themselves adequately for the future.

## **MYOCLONIC JERKS DURING FIRST TRIMESTER AS AN EARLY SONOGRAPHIC SYMPTOM OF ARTHROGRYPOSIS MULTIPLEX**

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**Ultrasound Unit, Department of Obstetrics and Gynecology, Neonatal Unit, Soroka University  
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Arthrogryposis multiplex is a multiple joint contraction syndrome. The etiology of this syndrome is hypokinesia/akinesia of joints resulting from a neurogenic disorder followed by myopathic disorder. Lately, a local Israeli syndrome was described as the lethal congenital contractural syndrome type 2 (LCCS2) (OMIM 607598) which is also associated with renal and cardiac malformations. A genome-wide linkage analysis, demonstrating linkage to approximately 6 cM homozygosity region on

chromosome 12q13 between markers D12S1604 and D12S83, was found. Therefore, early genetic detection of this syndrome is possible. However, many patients known to be carriers of this genes deny an invasive procedure and in others this genetic diagnosis is not found. Therefore, sonographic detection may be the sole tool for early detection of the syndrome. The sonographic appearance of LCCS2 in our area may appear late in second trimester. Therefore, early sonographic diagnosis may be important in counseling these patients, Lately, we have sonographically diagnosed 3 patients in our fetal malformation clinic at 13-14 weeks gestation. The main sonographic symptom was myoclonic jerks of hands and feet of these fetuses. Hands were flexed in the elbow joints and the jerks were from the shoulder girdle. Jerks were from the hip joint. At that time no joint contractures were apparent. These appeared only at 17 weeks gestation. In summary, tonic myoclonic jerks are demonstrated sonographically and can be an early neurologic sign for hypokinesia/akinesia syndrome. Future early sonographic observations should be performed in high risk patients prone to neurological accidents and congenital malformations.

## **THE GENETICS OF ARTHROGRYPOSIS MULTIPLEX CONGENITA**

**Vardiella Meiner, M.D.**

**Department of Human Genetics, Hadassah University Hospital, Jerusalem**

Arthrogryposis Multiplex Congenita (AMC) is a descriptive term which is used to describe a congenital appearance of joint contractures affecting at least two different areas of the body. AMC is clinically and genetically heterogeneous and includes over one hundred and fifty different conditions. All types of inheritance may be implicated, including chromosomal problems and the family tree may provide clues to how the condition is inherited.

Recently, following the identification of AMC-causing genes, several molecular mechanisms leading to arthrogryposis were elucidated leading to partial understanding of the muscular and neural pathophysiology of the disorder.

Early diagnosis and molecular analysis provides means for accurate genetic counseling and may in the future contribute to a more specific treatment.

## **NON-INVASIVE EVALUATION OF FETAL ANEMIA**

***Prof. Simcha Yagel, Dr. Tal Imbar***

**Hadassah Mt. Scopus Medical Center , Hebrew university, Jerusalem , Israel**

The in-utero course of the anemic fetus has improved dramatically, owing to early diagnosis and cordocentesis transfusion. In-utero invasive procedures such as amnio- and cordocentesis have become important modalities in the evaluation and treatment of anemic fetuses. However, they do carry risks for both the mother and fetus. A valid and sensitive non-invasive means of following the anemic fetus is evaluation of changes in middle cerebral artery peak systolic flow velocity (MCA-PSV). This is a sensitive tool for both the evaluation of fetal anemia and response to treatment. Intracerebral vessels respond earliest to the fetal anemic state, and are readily accessible for ultrasound examination.

We describe the non-invasive evaluation and management of the anemic fetus with MCA-PSV measurement, illustrated by index cases from our center.

MCA-PSV measurement is essential in the diagnosis, evaluation, and management of the anemic fetus. The use of this modality lessens the need for invasive procedures. The method is readily accessible and should be integrated into the repertoire of all obstetric ultrasound centers.

## **FETAL DETECTION OF CONUS MEDULLARIS**

**Oz Yuval, MD.**

**Obstetrics & Gynecology Ultrasound Unit,  
Rebecca Sieve Hospital SAFED**

Detection of fetal anomalies has broad spectrum issues of great importance. Central nervous system is one of the organs that our prenatal sonographic knowledge developed mostly during the last years. The progress of imaging equipment gave us the ability to demonstrate parts that we hardly saw before. Fetal neural tube defects are very important due to the possibility that concealed malformations may not be detected. Anomalies can be detected directly or indirectly. The value of direct identification is that it is not depending on the appearance of secondary signs. Demonstration of neural cord is important for identification of several defects. Demonstration of the conus medullaris looks to be important for evaluating spinal cord intactability and related defects. This screening presents the issue and the demonstration rate of conus medullaris during routine fetal organ screening.

### ***Session 5***

## **REHABILITATION OUTCOME OF CHILDREN AND ADULTS WITH MENINGOMYELOCELE (MMC)**

**Emmanuel Kornitzer, MD**

**Multidisciplinary Spina Bifida Clinic  
Alyn Pediatric & Adolescent Rehabilitation Center, Jerusalem**

Rehabilitation of children with MMC is directed mainly towards 4 aspects:

- Ambulation: teaching the children to walk with orthoses of various constructs. Children who do not achieve functional walking, need to acquire all the functional skills related to mobility in a wheelchair.
- Neurogenic bladder with incontinence of urine: the aim is to avoid urinary infections, dispose with the need for diapers and to be independent in CIC (clean intermittent catheterizations).
- Neurogenic bowel with incontinence of feces and/or constipation: the aim is to have regular voiding without soiling.
- Cognitive impairments (learning disabilities, organization skills, executive skills, etc): appropriate educational facilities and achieving all the skills needed to have a normal independent social life.

Success in achieving these goals is related to the neurologic functional level, cognitive development, motivation of child and family and a proper rehabilitation environment with daily training.

This presentation will detail our approach to these issues.

## **CASE PRESENTATIONS: Sessions 1-5**

### **Third Trimester Cerebellar Hemorrhage (S/1)**

**Hershkovitz Reli, Landau Daniella, Soroka Medical Center, Beer-Sheva, Israel**

Ultrasound Unit, Department of Obstetrics and Gynecology, Neonatal Unit, Soroka University Medical Center, Ben Gurion University of the Negev, Beer-Sheva

A 26 years old woman, married + one healthy child, was referred to the ultrasound (US) unit at 24 years gestation due to elevated maternal serum HCG of 4.37 MoM. Ultrasound examination revealed normal fetus and placenta. Karyotype was normal. The patient was followed at the US unit every 4 weeks. Fetal growth and Doppler blood flow velocimetry of the umbilical artery and uterine arteries were determined. At 36.4 weeks gestation, the patient was examined and sonographic examination demonstrated asymmetrical cerebellar hemispheres. The left cerebellar hemisphere was smaller than the right hemisphere. In its medial part, close to the vermis, a hypoechogenic area with thin septae was demonstrated. The first differential diagnosis was hemorrhage in the posterior fossa. In order to confirm the diagnosis MRI was performed. MRI examination confirmed the sonographic diagnosis. The couple was counseled and possible problems associated with the abnormal cerebellum were explained in the multidisciplinary clinic. They chose to terminate the pregnancy. A male neonate, after feticide, was delivered, without external malformations. Unfortunately, the couple refused post mortem evaluation of the neonate.

### **Fetal Delatation of the Small bowel (S/2):**

**Dr Hagit Nagar Pediatric Surgery, Dana Children's Hospital, Tel-Aviv**

A 34 y.o woman at 30 weeks gestation sought our medical advice. She was on a prenatal US follow-up b/o slight dilatation of the renal pelvis. US demonstrated marked dilatation of the small bowel. The couple was told that this picture is suggestive of midgut volvulus, which is known to be complicated by short bowel syndrome, and was advised to terminate the pregnancy. The same day they approached our team. Repeat US demonstrated dilatation of small bowel, with one loop extremely dilated crossing the fetal pelvis. Peristalsis proximal to the latter was slightly increased. Our presumptive diagnosis was ileal atresia, and we advised the couple to continue with the pregnancy. MRI supported the diagnosis. At 34 weeks' gestation, and 24 hours following hospitalizing for bleeding she gave birth to a baby girl birth weighing 2,150gr. Following adequate preparation 10 hours post delivery the baby was operated on. She found to have ileal atresia located at 7cm proximal to the ileocecal valve with old perforation and peritonitis. No intestinal malrotation was identified.

### **Suspected fetal right sided diaphragmatic hernia (S/2)**

**Dr Ron Rbinowitz and Dr. Ori Shen, Obstetric US Unit, Shaarei-Zedec Medical Center, Jerusalem**

A 27 year old women, gravida 2 , para 1, presented to our antenatal clinic complaining of vaginal bleeding, contractions and pressure at the end of the 38<sup>th</sup> week of gestation. The patient's obstetric history consisted of one , term , normal delivery. On the present pregnancy , Nuchal-Translucency, Triple test and Anomaly-Scan were not performed. On routine ultra-sound examinations on the 34<sup>th</sup> and 35<sup>th</sup> weeks of gestation , fetal growth restriction was suspected and polyhydramnios was observed. Ultra-sound examination at the time of admission revealed fetus in cephalic presentation , mild polyhydramnios, liver tissue and gal bladder occupying the right hemithorax with dilated liver vessels within the chest. pulmonary tissue was hardly noticed on the left hemithorax. No sign of placental abruption could be found.

With the diagnosis of right sided diaphragmatic hernia with liver tissue in the chest , she was transferred to the labor ward and delivered soon after.

QUESTIONS:

1. Is there any need for other methods of imaging ?
2. Is it recommended to deliver in the short term or wait natural delivery starts? spontaneously?
3. mode of delivery: cesarean section versus vaginal delivery or may be , exit procedure ?

**Fetal Ventriculomegaly with vermian and cerebellar hypoplasia (S/3):**

**Dr Dvora Kideron, Pediatric Pathology, Meir Hospital, Kfar Saba, Israel**

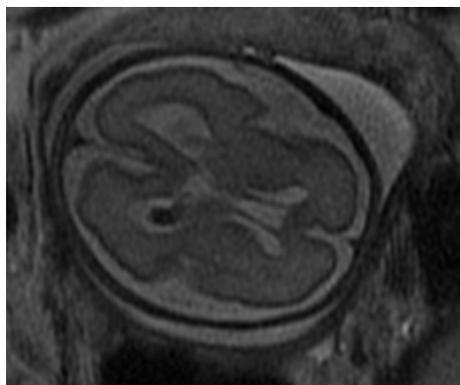
The patient is 33 years old, G2P. Routine fetal ultrasound screening in 14<sup>th</sup> week was normal. At 21 weeks, ventricular dilatation (16mm and 17mm) was noted, together with thin cerebral parenchyma. The 4<sup>th</sup> ventricle was dilated, with hypoplasia of cerebellum and vermis. Suspicion of postaxial polydactily was raised. The pregnancy was terminated on the 22th week. At autopsy, lissencephaly type II was found. Diagnosis of Walker Warburg syndrome was suggested.

**Featl intracranial bleed (S/3):**

**Dr. Lidia Gabis, MD, Chen Hoffman MD and Gad Barkai MD**

**Departments of Pediatrics, Radiology and Gynecology, Sheba (Tel Hashomer), Israel**

At 25 weeks the brain of a fetus showed a hyperechoic lesion in the left lateral ventricle, mild ventriculomegaly and haziness of the cerebellum. Previous ultrasound examinations were normal. He underwent an additional US after two weeks that showed the same findings and this prompted an MRI that was done at 27-28 weeks. MRI demonstrated mild ventriculomegaly (12 mm) without intraventricular hemorrhage, a choroid plexus T2-hypointensity and normal brain and cerebellum. The parents were advised that the fetus had an extracerebral bleed causing mild ventriculomegaly and if hydrocephalus will not evolve on follow up, the outcome should be uneventful. Laboratory examinations for TORCH and thrombastenia were performed. A follow-up MRI was done at 30 weeks and normal cortical and cerebellar development was demonstrated with minimal increase in the size of ventriculomegaly and choroid plexus lesion. The infant was born at 37 weeks and mild ventriculomegaly was demonstrated post-partum. No lesion of choroid plexus was shown. Subsequent US at 3 months showed the same. The baby's follow-up until age of one year showed normal development and head circumference.



**Twin to twin transfusion syndrome (TTTS) and ANEMIA of the fetuses – flipping roles after laser treatment (S/4):**

**Dr Nili Yanai, Gyn US Unit, Hadassah Medical center & Hebrew University, Jerusalem**

Twin pregnancy with diagnosis of TTS.

Laser therapy was performed.

Surprisingly, the DONOR baby (who initially supposed to be polycytemic) developed Anemia – this was reassured according to MCA Fetal US examination.

Diagnostic dilemma?

Treatment dilemma?

Blood transfusion was performed. Surprisingly, the recipient showed decreased MCA measurements with High Hematocrit. Therefore this time, Saline (NaCl) was transfused.

**Thoracic large MMC and long term treatment (S/5):**

**Dr. Dieter Class, Department of Neurosurgery, Katharinenhospital Stuttgart / Germany**

We report on a boy born April 2004 suffering from spina bifida with a large neural tube defect in the lower thoracic region. When the child was born he presented with hydrocephalus and a at this time small cyst in the cranio-cervical junction. **In addition we saw bladder exstrophy and anal atresia requiring surgical treatment. Further he had duplex kidney with vesicoureteral reflux and an atrial septal defect.**

1. **Any doubts about treatment?**
2. **What should be done to minimize morbidity?**

Neurosurgical treatment resulted in closure of the neural tube defect and implantation of a shunt system. 3 months ago we saw massive brain stem compression due to dramatic enlargement of the arachnoid cyst in the craniocervical junction requiring surgery again. After some days the shunt system failed due to obstruction of the central catheter requiring revision.

3. **Any thought to stop here or treat like any other kid with acute problem?**

Now the child is stable. He spent much time of his life in different hospitals. At no time any questions about the sense of treatment in this child with multiple handicaps were discussed. Once a child with spina bifida is born we aim to solve the problems coming up according to the necessities and the condition of the child on the other hand.

It is our policy not to enter into any discussion about what is called “quality of life”. We keep close contact to the parents to be prepared for new decisions to be made taking into account the neurological and pediatric development of the child.

**A BABY WITH GASTROSCHYSIS AND MENINGOCYSTOCELE – dilemmas of treatment (S/5)**

**Dr Yoram Bental, Neonatology, Laniado Medical Center, Netanya, Israel**

Baby delivered by C/S at 39 weeks gestation.

Birth weight – 3000 gr.

Parents - Nonconsanguinous Ashkenazi Jews.

7 healthy children at home.

No history of miscarriages.

Normal pregnancy.

On prenatal ultrasound MMC and Gastroschysis were suspected with enlargement of lateral ventricles on cranial U/S.

After delivery the baby was noted to have OEIS complex: Omphalocele, Extrophy of bladder, Imperforated anus and spinal defects.

Details of findings below.

The baby also has lower limb anomalies with poor movement of limbs below knee.

Cardiac echo – within normal limits

Eye exam – normal

Head ultrasound – mild enlargement of lateral ventricles.

Renal ultrasound – Lt kidney bigger than Rt., with very mild hydronephrosis 1.4 mm.

Chromosomes: 46, XY.

As the baby had an imperforated anus without any other opening of the small intestine, ileostomy was performed and the baby is being fed since.

Due to abnormal mechanical sucking the baby is fed orally and via a nasogastric tube.

Baby is alert, normal responses, normal neurological examination of upper part of body.

## הגישה האורולוגית לטיפול בילודים עם MMC ד"ר קובי בן חיים, אורולוגיה ילדים, מרכז רפואי ע"ש סוראסקי ובי"ח דנה לילדים, תל-אביב

MMC הינו הגורם השכיח ביותר לכיס שתן נירוגני בילדים. מצב זה מתבטא לעיתים בעצירת שתן כרונית – שאריות שתן גדולות ודלף שתן בטיפטוף ובמצבי עליית לחץ תוך בטני כגון: בכי. בהעדר טיפול מתאים הילדים עלולים לסבול מזיהומים חוזרים בדרכי השתן, מהופעה או החמרה של הדרואטרונפרוזיס ורפלוקס וסיקואורטרלי. בנוסף ללא טיפול במקרים רבים הילדים לא ישיגו שליטה במתן השתן ובצואה ולא יגמלו מחיתול בגיל הגמילה. מטרת הברור להעריך את תפקוד השלפוחית ודרכי השתן העליונים, לכן עליו לכלול אולטראסאונד דרכי השתן כולל מדידת שארית שתן (שצריכה להיות פחות מ-5 CC) בדיקת שתן לכללית ותרבית ותפקודי כיליה מייד לאחר הלידה. הבדיקה האורודינמית וציסטוגרפיה מבוצעות רק לאחר החלמה מהניתוח הנירוכירורגי. בתינוקות אשר לא מרוקנים את השלפוחית לאחר השתנה ספונטנית ניתן להשתמש ב-CREDE שיגרתית רק במידה ויעיל בריקון כיס השתן, ולא, יש להתחיל בצינתור לסירוגין על ידי ההורים X 4-3 ביום גם לפני ביצוע האורודינמיקה. טיפול זה יבטיח ריקון יעיל של כיס השתן והורדת הלחצים בתוכו ויקטין את הסיכון לפגיעה כליתית, זיהומים פיאלונפריטיים ונזק לדופן השלפוחית.

## שסע חד צדדי בשפה ושחלוף כרומוזמי בלתי מאוזן בעובר

ד"ר אמיר פלג, המכון לגנטיקה. מרכז רפואי "העמק" עפולה

שסע חד צדדי בשפה (unilateral cleft lip) נובע מכשלון באיחוי ה-maxillary prominence והרקע הינו רב medial nasal prominence בצד הפגוע. בכ- 97% מהמקרים מדובר במצב שאינו תסמיני והרקע הינו רב גורמי: שילוב של גורמים סביבתיים וגנטיים. בכ- 3% מהמקרים המום מופיע כחלק מתסמונת גנטית רחבה. תוארו יותר מ-200 תסמונות גנטיות שונות הכוללות שסע בצד השפה, בשילוב מומים נוספים או בעיות רפואיות מולדות אחרות. אין אחידות דעים בספרות לגבי הצורך בבדיקת כרומוזמים כאשר מדובר בממצא יחיד בבדיקת אולטרה-סאונד בהריון. במספר פרסומים דווח על סיכון מוגבר לליקוי כרומוזמי, בעוד שבפרסומים אחרים דווח כי סיכון זה אינו מוגבר מעל סיכון הרקע. אנו מציגים הריון עם שסע חד צדדי שאובחן כממצא יחיד, אצל עובר בשבוע 28 להריון. בבדיקת הכרומוזמים באמניוציטים נמצא קריוטיפ בלתי מאוזן, 47,XX,+marker chromosome. המשך הבירור הכרומוסומלי במשפחה גילה בבדיקת לימפוציטים של האם שחלוף מאוזן 46,XX,t(13;4)(q12;q35). בעזרת המידע אודות הקריוטיפ של האם, אובחן המרקר הכרומוסומלי אצל העובר 47,XX,+t(13;4)(q12;q35), והוא ביטוי של השחלוף בצורתו הבלתי מאוזנת שעבר בתורשה מהאם. שחלופים אוטוזומים בלתי מאוזנים קשורות בסיכון משמעותי לבעיות רפואיות שונות כולל מומים או פיגור שכלי. ההריון הופסק בשבוע 34 ע"פ בקשת בני הזוג. הממצאים לאחר הלידה אישרו את האבחנה הסונוגרפית ללא עדות למומים מבניים אחרים. המהלך בהריון שהוצג מצטרף לדיווחים בספרות הרפואית המצביעים על קשר בין סיכון מוגבר לליקוי כרומוסומלי לבין הדגמת שפה שסועה בהריון, גם כממצא יחיד.