SESSION I:

O1.

CONTRIBUTION OF MR IN THE DIAGNOSIS OF FETAL VENTRICULOMEGALY AND POSTNATAL FOLLOW-UP

Dr Catherine Garel, Fetal and Pediatric Radiology, France.

Fetal ventriculomegaly is diagnosed by US which can also define the type of ventriculomegaly (involvement of the frontal horns, the 3rd and the 4th ventricle) and look for extracerebral abnormalities. US can also look for some associated cerebral abnormalities such as callosal agenesis, haemorrhage, subependymal cysts …..
In our series of 326 ventriculomegalies, an associated cerebral abnormality was found in 41 % of the cases. 123 MR were performed in fetuses presenting an isolated ventriculomegaly in US. In 13,6 % of the cases, MR revealed associated cerebral abnormalities, which were overlooked by US : subependymal heterotopias, germinolysis, signal abnormalities of the white matter.
In the postnatal follow-up, MR was performed at the age of 2 months in 67 infants and 2 years in 26 children.
At 2 months, ventriculomegaly was observed in 46 infants (68 %). MR found additional findings in 8 infants (12 %): aqueductal stenosis (n=1), triventricular dilatation (n=2), ruptured septum pellucidum (n=1), subependymal abnormalities (n=1), thin corpus callosum (n=1), germinolysis (n=2).
At 2 years, ventriculomegaly was observed in 22 children (84 %). MR found abnormalities of the white matter in 10 children (6/ 10 are doing well) and arachnoid cysts in 3 children.

O2.

IS IT POSSIBLE TO VISUALIZE WHITE MATTER DISEASES ON FETAL MRI?

Dr Liat Ben-Sira, Ped Radiology, Dana Children's Hospital, TASMC (ICHILOV), Tel-Aviv[1]
Elka Miller[1], Dvora Kidron (Ped Pathology, Meir, Kfar Saba), Gadi Malinger (Gyn&Obs, Wolfson)

Prenatal MRI demonstrates high signal intensity in the white matter, that changes with time in the developing fetus. It is important to determine those with suspected brain damage and differentiate the normal white matter from the abnormal tissue.

Some examples including CMV infection, suspected metabolic disorders, ischemic and encephaloclastic abnormalities will be shown.

Further studies with DWI (Diffusion Weighted Imaging) in the fetus may further characterize white matter abnormalities

O3.

THE SPECTRUM OF CONGENITAL OBSTRUCTIVE VERSUS COMMUNICATING HYDROCEPHALUS.

Dr Liana Beni-Adani, Pediatric Neurosurgery, Dana Children's Hospital TASMC (ICHILOV), Tel-Aviv

The classification of congenital hydrocephalus (HCP) has changed since prenatal imaging became available, first with US and in later years – with Fetal MR. Thus, a baby born with hydrocephalus may present either a primary condition with malformative brain in association with progressive ventriculomegaly (VM), a "simple" mechanical obstructive problem leading to HCP (such as aqueductal stenosis), or aquired-in-utero (AIU) conditions with Fetal bleed or infection in an originally normal fetus.

The difference from classification in adulthood is apparent in two essential aspects of HCP: the disparity in the source pathologies that produce the HCP, and the practical distinctions in prognosis and treatment choices. No matter the underlying reason for HCP in the adult, most conditions can be divided to obstructive versus communicating HCP, which is not the case in fetal and neonatal conditions, where obstructive and absorptive hydrocephalic components may co-exist within the same patient.

Since malformative conditions develop during fetal life, in some cases very early during the first trimester of pregnancy, the duration and progressiveness of ventriculomegaly in-utero may cause irreversible secondary damage to CSF pathways, leading to shunt dependency for life. The long duration of HCP in-utero may also interfere with the ability of arachnoid granulations to absorb CSF, damage the cilia of ependyma in the ventricles (as has been shown in animal models of congenital HCP). In some infants the absorptive problem is transient, while in other infants it is a persistent problem that will need constant CSF extra-cranial drainage. It is great challenge for the pediatric neurosurgeon to distinguish between the different categories since, communication HCP was traditionally treated with shunt procedures, while obstructive HCP can be treated with endoscopic procedures either to avoid shunts at all or to minimize the numbers of shunts in a baby with multi-compartmental HCP.

O4.

MOLECULAR GENETICS OF CENTRAL NERVOUS MALFORMATIONS

Prof. Zvi U. Borochowitz, The Simon Winter Institute for Human Genetics
Bnai-Zion Medical Center, Technion-Rappaport Faculty of Medicine
Haifa, mdzvi@technion.ac.il

The recent remarkable advancement in the understanding of clinical genetics have shed light on the correlation between the genotype (genetic cause) and the phenotype (clinical presentation) of many disorders, among which are the human brain disorders. The understanding of the genetics of human brain malformations has led to insight into the formation of brain and into mechanisms of disease affecting brain. The brain seems to be a nonsegmented organ that is, however, formed in a segmented fashion by the overlap of genes that define anatomic and in all likelihood functional components of brain. Other genes and their encoded proteins regulate the processes of cell proliferation and migration; many of these genes have been identified based upon discoveries of human and mouse disease-causing genes. This insight has allowed one to consider a classification of these disorders by the genetic disruption. The past clinical-phenotypic based classification of these human brain malformations is thus been altered into a more genotype correlated classification based on the patterns of genetic expression.

The genetic programming of the neural tube may be summarized as a series of several principles. The expression of the organizer genes in gradients along the three principal axes of the neural tube is primordial. Amongst the most important concepts in its relevance to the

induction of abnormal neural development are: A)Upregulation of genes acting in the vertical axis (dorsoventral or ventrodorsal) causes hyperplasia and/or duplication of structures, and suppresses the expression of antagonistic genes; B)Downregulation of genes acting in the vertical axis causes hypoplasia or noncleavage (apparent midline "fusion") of structures; C)Upregulation of genes in the longitudinal axis causes ectopic expression in neuromeres where these genes are not normally expressed or alters the formation of structures in a most rostral or more caudal position than they normally occur; D)Downregulation of genes in the longitudinal axis results in hypoplasia of midline structures or deletion of entire neuromeres; E)Abnormal expression of genes of cellular lineage result in hamartomas with defective cellular growth and differentiation in addition to defective tissue architecture or disorganized arrangement of essentially normal cells.

The classification of human nervous system malformations as patterns of genetic expression is divided into several sub-groups: 1. genetic mutations expressed in the primitive streak or node, such as the EMX2 gene causing Schizencephaly; 2. disorders of ventralizing gradient in the neural tube such as SHH at 7q36 and PTCH at 9q22.3 causing Holoprocencephaly; 3. disorders of dorsalizing gradient of the neural tube, such as ZIC2 at 13q32 causing Holoprocencephaly; 4. disorders of the rostrocaudal gradient and/or segmentation such as EMX1 causing agenesis of the basal telencephalic nuclei; 5. aberrations in cell lineages by genetic mutation such as TSC1-hamartin, and TSC2-tuberin genes causing Tuberous Sclerosis; 6. disorders of secretory molecules and genes that mediate migrations such as Filamin-1 causing X-linked dominant periventricular nodular heterotopia; Doublecortin (DCX) causing X-linked dominant subcortical laminar heterotopia; LIS1, casing type I lissencephaly or Miller-Dieker syndrome; Fukutin, causing type II lissencephaly (Fukuyama muscular
dystrophy; 7. disorders of secretory molecules and genes that attract or repel axonal growth cones such as Netrin; and 8. disorders of symmetry such in Proteus syndrome were an apparent gene have been suggested-PTEN.

Despite such advancement, some of these malformations represent well-defined clinical and genetic entities for which there are specific tests; others have ill-defined genetic causes, while others can have both genetic and destructive causes. As such, the human brain malformations still represent clinical challenges for diagnosing and portends severe developmental disabilities and epilepsy recalcitrant to standard therapies.

The principle purpose of antenatal screening programmes is to identify diseases and than give the parents the best possible tools for them to decide on the continuation of the pregnancy, in the event of an affected fetus. Prenatal ultrasound screening tests reflects in most cases the walking of a blind in a 'Tigers Land', and the ability of this tool to determine a diagnosis is quit limited. The detection of a malformation dose not by itself permit the determination of it being part of a syndrome or of a non-syndromic option, and should therefore be considered non-specific, and are heterogenous and of non diagnostic possibilities. However, even though "Primum-non-nocere" is our main aim, with the present public attitude of litigations of "wrongful-life", deliver any prenatal sonographic changes to the family remains the only way. From the time that a higher risk was prenatally detected, and it was delivered to the couple, the basic bio-medical principles should be implemented, i.e., Primum-non-nocere; keep it in a non-directive Genetic Counseling manner; make sure the couple maintains a free reproductive choice; and make sure to eliminate all outside pressures.

The progress made in the understanding of the genetics of human brain malformations has led to insight into the formation of brain and into mechanisms of disease affecting brain. It bears upon clinicians, especially neurologists and geneticists to recognize the patterns of diseases of brain formation, to properly diagnose such disorders, to assist in the recurrence risk of these malformations and to guide families with appropriate expectations for outcomes.

SESSION II:

O5.

NORMAL DEVELOPMENT AND PATHOLOGY OF THE POSTERIOR FOSSA

Dr Catherine Garel, Fetal and Pediatric Radiology, France.

Anatomical and embryological data of the development of the posterior fossa are reminded.
The normal appearance of the different structures of the posterior fossa is described in US and MR.
The contribution of both imaging modalities in the different pathologies is discussed.

- Malformations : Dandy-Walker malformation, vermian agenesis, cerebellar hypoplasia, rhombencephalosynapsis.
- In utero acquired pathologies : ischaemia, haemorrhage, infectious disease, tumors and arachnoid cysts.

The main advantage of MR over US is to provide a better visualisation of the vermis and the brain stem independently of the position of the fetal head and the thickness of the maternal abdominal wall. It is more sensitive than US in the detection of ischaemia or haemorrhage.
However the spatial resolution is not good enough to be sure that the vermis is complete so that an accurate diagnosis of some malformations remains difficult in the antenatal period.

O6.

FETAL STROKE – GRADE MATTERS

Uriel Elchalal, Simcha Yagel, John Moshe Gomori, Shay Porat, Liana Beni-Adani, Nili Yanai, Michel Nadjari

Objective: To determine if the severity of antenatally diagnosed hemorrhagic fetal brain lesions (fetal stroke) detected by ultrasound and magnetic resonance imaging (MRI) predicts postnatal neurodevelopmental prognosis.
Study design: In-utero presentation and postnatal neurodevelopmental outcome of sonographically detected subdural hematoma or fetal stroke presenting as intraventricular hemorrhage (IVH) or intraparenchymal brain hemorrhage, are described.
Results: Of 33 fetuses diagnosed with hemorrhagic brain lesions, 17 were electively terminated and 2 suffered intra-uterine fetal demise (IUFD). Thirteen were live born, seven by cesarean delivery and six by spontaneous vaginal delivery. One case was lost to follow-up. Eight neonates had moderate to severe neurological deficit by a mean age of 35 months (range 6-96 months). One died at two months of age. These nine were diagnosed with grade III-IV IVH *in utero*. Four neonates had normal neurological outcome by a mean age of 41 months (range 30-48 months); these four were diagnosed with subdural hematoma (n=1) or grade I-II IVH (n=3) *in utero*. Fourteen cases were followed up with MRI, which confirmed ultrasound findings in ten (71%). In three cases (21%) MRI diagnosis was more accurate and grading more severe than ultrasound. Unilateral left hemispheric lesions were much more common than right sided lesions (13 vs 1 respectively).
Conclusions: An antenatal sonographic diagnosis of fetal stroke with IVH grade III-IV or with brain parenchymal involvement appears to be associated with poor neurological outcome. MRI may contribute to the accuracy and increase severity of diagnosis, particularly in grade II and III lesions. Left-sided unilateral lesions are more common than right.

O7.

QUANTITATIVE MRI MEASUREMENTS OF HUMAN FETAL BRAIN DEVELOPMENT *IN UTERO*

R. Grossman, Neurosurgery Dept.; C. Hoffman, Radiology Dept., Y. Mardor Advanced Technology Dept. and A. Biegon, J. Sagol Neuroscience Center, Sheba Health Center, Tel Hashomer.

Magnetic resonance imaging (MRI) allows for high resolution imaging of the central nervous system. We have tested the feasibility of using MRI in conjunction with quantitative image analysis to perform volumetric measurements of the brain in the developing human fetus *in utero*. The data base comprises a total of 50 fetuses (gestational age 22-41 weeks) referred because of suspected infarct, hydrocephalus, ventricular asymmetry or enlargement, or risk of abnormalities due to family history or maternal illness. Scans were obtained using a 1.5T magnet and a single shot fast spin echo (SSFSE) T2 sequence, slice thickness of 3mm, no gap and analyzed with NIH Image software. All scans were evaluated by an expert neuroradiologist.. The intra-rater error was 5%-9% for 2 raters, while the inter-rater error was 13%. Therefore, a single rater then performed all the measurements. The right and left hemisphere and lateral ventricles were identified and their area measured on all relevant slices. The ratio (%) of ventricles to hemispheric parenchyma was significantly correlated with developmental age (gestational week) in normal fetuses ($N=26$, $R=0.61$, $p<0.001$) and very sensitive to ventricular pathology. The mean values in normal fetuses was 5.4% (SE 0.66) compared to 37.4% (SE 17, N=6) in fetuses with ventriculomegaly. These preliminary results support the use of image analysis and MRI to produce quantitative severity assessments of brain pathologies in the developing human fetus.

O8.

PRENATAL ULTRASONOGRAPHIC DETECTION OF URINARY TRACT ANOMALIES: AN INTERDISCIPLINARY TEAM APPROACH.

Tzipora Falik-Zaccai ,MD,Nechama Kfir, M.S.c Nasser Wael,MD, Irith Weismann, MD.
Division of Medical Genetics, Division of Pediatric Nephrology
Western Galilee Hospital Naharia, Rappaport Faculty of Medicine, Technion, Haifa.

Detection of congenital urinary tract anomalies (UTA) in prenatal ultrasound accounts for about 20% of all abnormalities found on scanning. Findings might be benign and reversible or indicative of significant medical problems, associated with extra-renal defects, or component familial syndromes.

Thirty-nine couples were referred to the nephrology - genetic clinic. Nine approached the clinic due to known UTA or genetic nephrological diseases. Thirty-one were after 2nd level ultrasonographic evaluation with UTA identified. Family history and interpretation of the abnormal ultrasonographic findings detected in their fetus were provided. Genetic workup and follow up during the pregnancy and following delivery were recommended to avoid unnecessary terminations of pregnancy and to assure post-natal evaluation and treatment of the newborn. We identified 8 families at high risk for primary hyperoxaluria, cystinosis , janssen syndrome, congenital nephrotic syndrome, multicystic dysplastic kidneys and horseshoe kidney and familial reflux nephropathy. Additional 31 presented with fetal uni/ bilateral hydronephrosis, renal cysts, horseshoe/ectopic kidneys and single kidney. Five pregnancies were terminated. Thirty-four continued to term and the ultrasonographic diagnosis confirmed in 31. All newborns were followed in the genetic nephrology clinic. Informative decisions regarding the management and outcome of pregnancies with possible UTA or hereditary kidney disease, couples should be counseled regarding the clinical and genetic applications involved. Early knowledge of the abnormality gives the opportunity to treat as early as possible the neonate without aggravating the clinical situation due to delayed therapy. Close co-operation between ultrasonographers, pediatric nephrologists and medical geneticists is extremely beneficial for couples at risk.

O9.

THE APPROACH TO FETUSES WITH MICROCEPHALY IN THE MULTIDISCIPLINARY FETAL NEUROLOGY CLINIC.

Dorit Lev, Tally Lerman-Sagie, Gustavo Malinger.
Fetal Neurology Clinic, Edith Wolfson Medical Center

Microcephaly is defined postnatally as low brain weight and a small head circumference (HC) more than two standard deviations (SD) below the mean or below the 3rd percentile. Such a broad definition obviously includes normal individuals. The smaller the head circumference, the higher the chances of associated mental retardation. Prenatally, there is no consensus regarding the exact definition of abnormally small HC, some authors propose the -2SD cutoff while others propose the -3SD cutoff. Most fetuses with relatively small head circumference measurements, (around the 3rd centile) have in fact an "intrauterine growth restriction" and will grow regularly throughout gestation and eventually turn out to be normal babies.
The prenatal diagnosis of microcephaly, particularly in cases of primary microcephaly, is usually difficult before the 3rd trimester**.** In some cases the presence of microcephaly may be suspected based on additional sonographic findings.
Primary microcephaly is genetically heterogeneous, with several loci currently mapped. The recurrence risk for parents of a child with primary microcephaly is 25%.
The etiologic heterogeneity and variability of microcephaly in genetic syndromes are among the more difficult issues in prenatal ultrasound in pregnancies either with an incidental finding of this anomaly, or in cases with a recurrence risk**.**
The counseling for fetuses with a small HC is difficult. Mental retardation can safely be predicted in cases with associated US findings, abnormal karyotype or positive test for intrauterine infection. In fetuses with isolated small HC an effort should be made to determine gyral normality in utero by US or MRI.
To illustrate these difficulties we will present our experience with fetuses followed for microcephaly in our clinic, including neuro-developmental follow-up of these babies. A flow chart for prenatal investigation of fetuses with microcephaly will be displayed.

O10.

THE ROLE OF THE MEDICAL GENETICIST IN THE MANAGEMENT OF THE FETUS WITH MALFORMATIONS: THE CONGENITAL HEART DISEASE EXPERIENCE

Annick Raas-Rothschild, Nadjari Michael, Yagel Simha, Yanai Nili, Michal Becker-Cohen, Vardiella Meiner, Michal Sagi, Sagui Gavri, Zeev Perles, Azaria JJT Rein.
This is a time of unprecedented increase in knowledge on the genetic basis of disease while the technology is rapidly changing. Genetic services have been defined as health measures implemented to help people with a genetic disadvantage and their families to live and reproduce as normally as possible. They can be divided into those services, which target whole populations, and services, which focus on the needs of families, which are affected by a genetic disorder.
The prenatal consultation includes the geneticist and the pediatric cardiologist counseling. The likelihood of the congenital heart disease (CHD) being a part of a syndrome or connected to other genetic disease is investigated. In parallel or at the same clinic (cardio-genetic clinic), comprehensive fetal echocardiography and explanation of the cardiac findings to the parents including the postnatal clinical implications of the fetus CHD are offered.
The cardio-genetic experience for pre- and postnatal counseling to the family of a child affected by a congenital heart disease will be presented.
Teamwork and communication is necessary between obstetrician, pediatricians, geneticist and ultrasonographer to explain as objectively as possible the significance of any malformation to the pregnant couple.

O11.

TERMINATION OF PREGNANCY FOR MAJOR FETAL MALFORMATIONS IN THE NEGEV POPULATION DURING YEAR 2003

Gilad Bodenheimer Medical student (MD thesis), Dr. Reli Hershkovits, Head of the ultrasound unit(Ob/Gyn dept), Dr. Daniela Landau, Head of newborn dept,
Dr. Dahlia Weitzman, Epidemiology and health services evaluation
Soroka medical center, and the Faculty for health science, Ben-Gurion University of the Negev

Objectives: This study was performed in order to survey pregnancies in which a major fetal malformation was found, and to determine the factors leading the pregnant women to terminate their pregnancy before and after 23 weeks of gestation in the two ethnic populations of the Negev.

Study design : Data collection was performed retrospectively from the medical records of 174 women with at least one major fetal malformation. The diagnosis was established by the tertiary ultrasound clinic and genetic institute in our hospital during the year 2003. Statistical analysis included univariate and multiple logistic regression.

Results: In 2003, 55.3% of livebirths in the Soroka Medical Center were Bedouins and 44.7% Jewish. 53.4% of major fetal malformations were found among Bedouin women as opposed to 46.6% in Jewish women. Early gestational age at diagnosis was found to be related to past termination of pregnancy, older women, and two or more previous pregnancies with fetal malformation among Bedouin women. The low rate of termination of pregnancy among the Bedouin women (33.7% vs 61.7% among Jewish women), along with the fact that the major fetal malformations were more severe among the Bedouin women may explain the higher perinatal motality rate from congenital malformations (31.5% vs 11.1% among Jewish women). 56% of pregnancies with fetal malformations were diagnosed after 23 weeks gestation in Bedouin women (vs 44.4% between 18-13 weeks gestation in Jewish women). 30.9% (25 cases) of the major malformations diagnosed after 23 weeks of gestation could have been diagnosed earlier. The most significant predictor for termination of pregnancy at any time was woman of Jewish origin vs Bedouin origin.

Conclusions: These findings point the importance of early diagnosis and it emphasizes the need to investigate and understand the causes for early or late diagnosis. It points out the necessity to treat differently the different populations, due to religious and cultural differences.

O12.

COMMISSURAL ANOMALIES – THE EXPERIENCE OF THE FETAL NEUROLOGY CLINIC

G. Malinger, D. Kidron, L. Ben-Sira, D. Lev, T. Lerman-Sagie

Wolfson Medical Center, Holon, Sapir Medical Center, Kfar Saba, Dana Children Hospital, Tel-Aviv

Sackler School of Medicine, Tel Aviv University, Tel Aviv, Israel

Objective To describe the imaging and clinical findings in a cohort of fetuses with commissural anomalies (CA).

Methods We reviewed retrospectively, the files of all the fetuses diagnosed as affected by any type of CA during the period of 1989-2005.

Results Fifty-five fetuses with CA were identified. Only 8 cases were diagnosed during 1989-1999, the remaining 47 were diagnosed in the last 5 years. The mean gestational age at the time of US diagnosis was 26.8 weeks (range 15-38 weeks). Complete agenesis of the corpus callosum (ACC) was present in 25 fetuses, partial ACC in 6, dysgenesis of the CC in 14, the CC was considered thick in 3, a lipoma of the CC was diagnosed in another 3, two fetuses had vellum interpositum cysts, one with ACC but normal anterior commissure and one with the middle interhemispheric variant of holoprosencephaly. CA were isolated in 21 patients; associated anomalies were found in the remaining 34. Colpocephaly was present in only 24 fetuses, 21 of them with complete ACC.

Associated anomalies were found in 11/25 fetuses with complete ACC and in 22/30 fetuses with other forms of CA.

MR imaging was performed starting form 2000 in 15 patients. Thirty-seven patients opted for termination of pregnancy, in one fetus IUFD occurred, 16 were delivered and one pregnancy is still ongoing. One child died close to delivery due to fetal akinesia syndrome and 2 were lost to follow-up.

Nine children have normal development (mean follow up 25.2 months). The remaining have: severe psychomotor retardation (1), mild developmental delay (2), optic nerve hypoplasia (1).

Conclusion Commissural anomalies can be diagnosed by US. The increasing knowledge of brain anatomy gained in the last years seems to be an important factor in the increased diagnostic yield. Following prenatal counseling, 67% of our population opted for TOP, this number is even higher (74%) if we excluded 5 fetuses with relatively benign findings (lipoma and cavum interpositum cyst). On the other hand, patients with the US and MRI diagnosis of isolated complete ACC or ACC lipomas seem to have a good prognosis (6/6).

Currently it is clear that fetuses with dysgenesis or partial agenesis of the CC have almost always associated anomalies, thus the prognosis in these cases is reserved. Continuation of pregnancy may be offered to parents of fetuses with isolated ACC.

O13.

THIRD VENTRICLE ENLARGEMENT IN INFANT WITH TRISOMY 21 - A NEW FINDING.

Michael S Schimmel, Cathy Hammerman, Ruben Bromiker and Itai Berger

Introduction

On routine head sonography of babies with trisomy 21 (T21) we noticed that the third ventricle (3V) seemed to be larger than expected.

Objective: To determine whether the size of the 3V, as observed on head ultrasound, is statistically different in babies with and without T21.

Methods

Routine head ultrasounds were performed in all term babies with T21 within seven days of birth and were compared to those of randomized term infants without T21. Studies were done with 7.5 probe through the anterior fontanel. Statistical significance was calculated by t test and defined by a $p<0.05$.

Results

65 babies were evaluated, 44 with T21 and 21 infants without T21. Although both groups were of similar gestational age (39±1 vs. 39±1 weeks), babies with T21 were generally smaller (3.076±0.365 vs. 3.375±0.399 kg; p=0.004) with smaller head circumferences (HC) (33±1.3 vs. 34.9±1.7 cm; p=0.001). Despite the smaller overall HC, both the width and the length of the 3V were ***enlarged*** in the T21 babies (see table below). For comparison, sizes of the lateral ventricles (similar in the two groups) are presented.

	Rt Lat Vent	Lt Lat Vent	3rd Vent Length	3rd Vent Width
T21 (n=44)	0.15+0.06	0.17+0.1	0.90+0.26	0.27+0.08
Ctrl (n=21)	0.13+0.04	0.15+0.05	0.63+0.3	0.19+0.06
Sig	0.18	0.4	0.001	0.001

Discussion

Our data present an isolated finding of enlarged 3V in infants with T21. When viewed in the context of a smaller overall HC in these infants and a lateral ventricle size that is similar to that of the control infants, this finding is even more intriguing in that it does not appear to represent a generalized brain atrophy. Further research should be done in order to assess possible maldevelopment of brain structures surrounding the 3V, as well as the clinical and prognostic implications of these findings.

O14.

NEUROLOGICAL OUTCOME OF CCA – A LITERATURE SURVEY AND CASE STORIES.

Dr Yael Leitner, Ped Neurology, *(Tel-Aviv, Dana children's Hospital)*

For an increasing number of fetal CNS anomalies, means of diagnosis have progressed faster than knowledge of the prognosis.

Agenesis of the corpus callosum (ACC), whose diagnosis can be verified through prenatal ultrasonography and MRI illustrates this problem well.

Agenesis of the corpus callosum, either complete or partial is a common brain malformation with a prevalence of 0.5- 70 per 10,000 and 230 per 10,000 in children with developmental delay.

ACC is usually associated with other CNS or extra- CNS malformations. It has also been described in many chromosomal anomalies, viral infections, metabolic diseases and toxic syndromes.

Common symptoms of ACC are mental retardation, epilepsy and/ or behavioral disorders.

In the past, most of the cases with ACC were discovered during the assessment of significant developmental delays or malformative syndromes.

Isolated ACC (without associated anomalies) seems to be rare, and was usually revealed as an incidental finding in brain imaging performed for other indication (e.g.: head trauma). Many of these patients were described as neurologically asymptomatic. Other cases of isolated ACC were discovered in adults undergoing investigation for epilepsy , or were revealed later in childhood by relatively minor symptoms like learning difficulties or by behavioral disorders.

Today- fetal screening by ultrasonography and by MRI have changed the picture since many cases of ACC, and more specifically cases of ***apparently isolated ACC***

(with normal karyotyping and no associated anomalies) are diagnosed in mid-gestation. Under these circumstances we are faced with the question of future prognosis. While most investigators agree that ACC associated with other anomalies carries a bad prognosis, several investigators have lately described that isolated ACC diagnosed prenatally carries a good prognosis with 85% normal outcome.

Some of the studies are retrospective, while other describe a short follow- up period, or partial cognitive assessments.

The lecture will briefly review the current literature concerning this issue, in an attempt to find out if prenatally diagnosed "asymptomatic" ACC does actually exist.

A few cases of prenatally diagnosed ACC will be presented.

O15.

- O15: Fetal management case: Dr Kidron, Ped Pathology *(Meir, Kfar saba)*. *15.05-15.25*

O16.

CAN FETAL MR HELP IN CHARACTERIZING SUSPECTED CYSTIC KIDNEY DISEASES IN THE FETUS

Liat Ben-Sira, Elka Miller, Miriam Davidovitch *
Division of Pediatric Radiology, Radiology Department.
Dana Children's Hospital, Tel Aviv, Sourasky Medical Center.
*Pediatric nephrology, Shneider Childrens' Hospital, Petach Tikva, Israel.

Fetal MR is an important complementary study to ultrasonography of fetal urinary tract anomalies.
Sixteen fetuses with suspected urinary tract abnormalities were referred to MR. Pathology included multicystic dysplastic kidney (5); other cystic diseases in sonographic echogenic kidneys (4), abnormal position of the kidneys (4) and obstructive uropathy (3).
Most cases of multicystic dysplastic kidney, abnormal position and obstructive uropathy, are straightforward, however; obstetricians are increasingly facing the challenge of counseling pregnant women with sonographic echogenic kidneys that carry an uncertain prognosis.
Hyperechoic kidneys represent a challenging diagnosis because of the wide variety of etiologies and uncertain outcome. MRI has an inherent ability for better tissue contrast differentiation and characterization and may offer an additional tool for better delineation of the parenchyma.
In two fetuses with a diffuse hyperintense parenchyma, we were able to demonstrate tiny hyperintense lesions on T2-weighted images that were not recognized on the initial ultrasound images and later proved to be tiny cysts. Thus we could establish the diagnosis of hereditary cystic disease.
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O17:

FETAL MR OF THE NORMAL AND PATHOLOGICAL GASTROINTESTINAL TRACT

Dr Catherine Garel, Fetal and Pediatric Radiology, France.

After a brief overview of fetal gastrointestinal (GI) tract physiology, the normal patterns of the GI tract are described. The main principles to understand MR abnormal findings are given. The results of a study in 24 fetuses are reported regarding the contribution of fetal MR in the evaluation of duodenojejunal obstruction, small bowel obstruction and pathology of the colon and rectum.
The conclusions are that MR is a good method to localize an obstruction, to diagnose a microcolon, to assess the normality of the colon and to localize a pelvic mass. It is sometimes valuable for diagnosis of anorectal malformation and rectourethral fistula. It is not a good tool to appreciate the amount of normal small bowel and to assess multiple obstructions.

O18.

FETAL OVARIAN CYST – THERAPEUTIC APPROACH

Hagith Nagar, Pediatric surgery, Dana Children's Hospital, TASMC, Tel-Aviv

Fetal ovarian cysts are usually found on prenatal routine ultrasound during the third trimester of pregnancy. The etiology is not entirely clear and there is a wide range of differential diagnosis. As the most important factor is the outcome, accurate diagnosis is of primary importance for a favorable outcome.
Intrauterine torsion is the most common complication. Torsion occurs more frequently in fetal life than postnatal. The causes for torsion are the length of the cyst pedicle and the diameter of the cyst. The first can only be found during surgery and the second has not been conclusively agreed upon. The accepted treatment is aspiration of cyst during pregnancy but this procedure is not free of complications.
Therapeutic criteria should be carefully weighed: cyst size, rapid growing cyst
A number of questions should be addressed. Is intrauterine aspiration a safe procedure? How accurate is the US in identification of torsion, ovarian cyst or cysts of other origins? Should delivery be induced? Following delivery, should the cyst be aspirated or surgically removed?
These and many other issues should be discussed.

O19.

FETAL LUNG DYSPLASIA: CLINICAL OUTCOME BASED ON NEW CLASSIFICATION SYSTEM

R ACHIRON[1], Y ZALEL[1], S LIPITZ[1], J HEGGESH[2], R MAZKERETH[3], J KUINT[3], J JACOBSON[4], S YAGEL[5]
Departments of Obstetrics and Gynecology[1], Pediatric Cardiology[2], Neonatal Intensive Care Unit[3] and Pediatric Radiology[4], Sheba Medical Centre, Tel-Hashomer

Objective: To evaluate the clinical application of a new classification system of fetal lung anomalies.
Methods: Forty fetal diagnoses of lung lesions were analyzed according to our proposed classification system, in which each lung component is considered using 2-D ultrasound and color and power Doppler technology. Medical files, natural history and neonatal follow-up were recorded.
Results: **Type I dysplasia:** 4 cases of agenesis of the lung were diagnosed, three with right lung agenesis and one with left lung agenesis. Three of the four patients elected to terminate the pregnancy. The surviving fetus was diagnosed with scimitar syndrome and postnatal embolization of the aberrant vessel was performed.
Type II dysplasia: 1 case of normal lung with abnormal systemic feeding artery was diagnosed with normal neonatal outcome.
Type III dysplasia: Abnormal lung with abnormal vascularity was found in 14 cases, presenting in most cases as echogenic lung masses. Seven were supradiaphragmatic, 6 subdiaphragmatic and one case was of undetermined situ. All 14 fetuses showed aberrant systemic artery emerging from the aorta.
Abnormal venous drainage could be identified in only 5 of the fetuses (35%): 3 had prominent azygos vein, one showed drainage to the IVC and one had multiple intrapulmonary veins forming a huge arterio-venous (A-V) shunt. Two cases in this group terminated the pregnancy, the case with A-V shunt following development of hydrops, and one at maternal request. All of the remaining 12 fetuses (86%) survived and are alive and well; only one of them needed immediate postnatal embolization of bilateral aberrant feeding arteries.
Type IV dysplasia: Abnormal lung with no vascular abnormality was diagnosed in 20 fetuses. In this group there was one case of IUFD, two patients terminated the pregnancy, one complicated with hydrops and one upon maternal request. Survival rate in this group was 85%. Only two cases needed immediate surgical repair.
Type V miscellaneous dysplasia: One fetus demonstrated echogenic lung with split notochord syndrome and survived.
Conclusions: Congenital bronchopulmonary and related vascular anomalies can be categorized using the new classification system. This new approach enabled prenatal evaluation of each lung component and facilitated cogent management of the fetus with congenital lung dysplasia.

O20.

THE DILEMMA OF ONE ABNORMAL IN MULTIPLE PREGNANCY.

Dr. Nili Yanai, Hadassah Ein Kerem, Jerusalem

One of the major dilemmas in managing multiple pregnancies is a single complicated fetus.
Several conditions will be discussed, regarding this issue.
The management of a single fetal death should consider the risk for the survivor and the maternal and pregnancy risk. What are the expecting conditions in relation to monochorionic versus dichorionic pregnancy. Above 60% of monozygotic twins are monochorionic, and about 15%-25% of them will be associated with twin to twin transfusion syndrome (TTTS), in which a single fetal death is not rare.
Discordant twins, results from an intrauterine growth restriction of one, leads to another dilemma. Dealing with the arrest of growth of a single pregnancy is commonly associated with premature delivery, but regarding twins pregnancy, remote from maturity, this management may harm the non affected fetus. The priorities, timing and the way of delivery will be dicussed.
Another issue in this context is a single malformed fetus. In this session one should consider the severity of the anomaly, the prognosis, mortality and morbidity of the affected fetus regarding the risk and the benefit of a selective termination.
Invasive procedures, diagnostic (such as amniocentesis, chorionic villi sampling) versus therapeutic (as selective termination or vascular ablation) should also be discussed, dealing with the tremendous dilemma of a complicated one of the multiple fetuses.

021.

- **O21: Ethics/law/ in Fetal Medicine – Rabi Y. Weiner:**

"What is the ethical aspect of risking the healthy fetus when the other fetus has a major congenital anomaly with bad prognosis":
1. in intervention for further diagnosis. 2. In fetoreduction