PPS 2012

58th Annual Meeting
Paediatric Pathology Society

Hotel Me Madrid - Reina Victoria
Plaza de Santa Ana

Madrid, Spain
October 25th - 27th
2012

www.pps2012madrid.com

Index

Introduction

It is our pleasure to welcome you to the 58th annual meeting of the Paediatric Pathology Society, in Madrid, Spain, from October 25th to 27th, 2012.

This is the largest paediatric pathology meeting in Europe and we have endeavoured to make it attractive. This year we are organising a Symposium on “Paediatric Dermatopathology”, with the participation of the most renowned paediatric dermatologists and dermatopathologists from Spain and Europe. The Slide Seminar on “Unexpected Causes of Perinatal Death” promises to be a success. At the oral presentations, we will be able to share our latest research topics and clinical practice with the leading paediatric pathologists. Finally, we will count on the presence of Professor Roger Byard, who will honour us with the John Emery Memorial Lecture.

This exciting event will take place at the Hotel ME Madrid Reina Victoria, a recently refurbished old and famous hotel located downtown, which used to host the world’s famous bullfighters. We have chosen this venue because of the beautiful architecture, the amazing views of the city and the modern meeting rooms and facilities. It has all the amenities that make it a perfect meeting place. For those interested in art and culture, there are plenty of museums, opera and theatres within walking distance. Here is where the PPS´s excellent scientific sessions will be held, ensuring the most enjoyable visit for you.

We are looking forward to welcoming you and we will do our utmost best to make your stay in Madrid truly unforgettable!

Yours sincerely,

Isabel Colmenero
Organizer

Fernando Casco
Co-organizer

Committees

Organizing Committee

Local Organizer	*Isabel Colmenero*
Co-organizer	*Fernando Casco*

Scientific Committee (Officers of the Society)

President	*Christina Vogt* (Norway)
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	Anastasia Konstantinidou (Greece)
	Maureen O'Sullivan (Ireland)
	Sergey Popov (Russia)
	Tamas Marton (UK)
	Marian Malone (UK) ex-officio
	Marta Cohen (UK) ex-officio
	Claire Evans (UK)
	Rita Alaggio (Italy)

Program

Thursday 25th

16:00-18:00	**Registration**
16:30-18:00	**Committee Meeting**
18:00-20:00	**Symposium: "Paediatric Dermatopathology"** Chairpersons: *Marian Malone* and *Antonio Torrelo*
18:00-18:30	"Spitz Nevus and Spitzoid Melanoma" *Luis Requena*, Spain
18:30-19:00	"Cutaneous lympho-haematopoietic proliferations in children" *Marie-Anne Brundler*, UK
19:00-19:30	"Cutaneous manifestations of autoinflammatory syndromes" *Antonio Torrelo*, Spain
19:30-20:00	"Paediatric skin tumors" *Marian Malone*, UK
20:30	**Welcome Cocktail at "Hotel Me Madrid Reina Victoria"**

Friday 26th

09:00-10:45 **Oral Presentations**
Chairpersons: *Ronald de Krijger* and *Aurore Coulomb*

09:00-09:15 **O01** - Synchronous pancreatic and colonic adenocarcinoma in a 16 y/o Inuit male with homozygous PMS2 DNA mismatch repair gene deficiency syndrome.
Camelia Stefanovici, Kris Milbrandt, Andrew McKay, Michael Cantor, Bernie Chodiker, Rochelle Yanofsky, Albert E. Chudley.

09:15-09:30 **O02** - Study of bax expression by RT-PCR from paraffin embedded pediatric astrocytomas correlates with lower survival in low grade.
Pedro Valencia Mayoral, Pilar Eguía Aguilar, Mario Pérezpeña-Díazconti, Fernando Chico Ponce de León, Miguel Ángel Méndez García, Francisco Arenas Huertero.

09:30-09:45 **O03** - Vascular abnormalities in intestinal atresia.
Pedro Valencia Mayoral, Guillermo Ramón García, Mario Perezpeña-Díazconti, Victor Haro Sánchez.

09:45-10:00 **O04** - Genetic analysis of Pleuropulmonary Blastoma (PPB) by CGH and FISH.
C. Vokuhl, L. de Leon, S. Kirsch, E. Koscielniak, I. Leuschner.

10:00-10:15 **O05** - Upregulation of HOXB4 plays a role in the pathogenesis of nephroblastomas and is mediated by miR-23a.
Karin Koller, Suman Das, Ivo Leuschner, Melanie Korbelius, Gerald Hoefler, Barbara Guertl.

10:15-10:30 **O06** - Two cases of Leiner's Disease.
Helen Wainwright, Roc Kashula.

10:30-10:45 **O07** - Prevalence of duodenal bulb involvement in Coeliac Disease.
Sonja Mansfield-Smith; Nagendra Rao, Mike Thomson, Marta C. Cohen.

10:45-11:15 **Coffee Break**

11:15-12:00	**Oral Presentation** Chairpersons: *Marie-Anne Brundler* and *Fernando Casco*
11:15-11:30	**O08** - Type 2 Mosaicism of Nevoid Basal Cell Carcinoma Syndrome. *Isabel Colmenero, Fernando Casco, Ángela Hernández-Martín, Antonio Torrelo, Luis Requena.*
11:30-11:45	**O09** - CLOVES and Klippel-Trenaunay Syndromes Share Somatic Mosaic Activating Mutations in PIK3CA. *Valerie L. Luks, Ahmad I. Alomari, Steven J. Fishman, Samantha A. Spencer, John B. Mulliken, Harry P.W. Kozakewich,Matthew L. Warman, Kyle C. Kurek*
11:45-12:00	**O10** - Splenic cysts associated with hematuria. *G. Guarda Muratori, C. Otal Salaverri, A.García Escudero, N. Conde Cuevas, J. Armas Padrón, I. Ortega Medina, P. Gallinato Pérez, R. González Cámpora*
12:00-13:00	**John Emery Memorial Lecture: Prof. Roger Byard** Chairperson: *Christina Vogt*
13:00-14:30	**Lunch Break**
14:30-16:30	**Symposium "Perinatal Pathology"** Chairpersons: *Anastasia Konstantinidou* and *Peter Nikkels*
14:30-15:00	"Post mortem examination of very small fetuses. How useful is it?". *Cesar Peres,* UK
15:00-15:30	"Placental pathology and adverse perinatal outcome" *Peter Nikkels,* The Netherlands
15:30-16:00	"What is in a rib? Examination of the rib in perinatal and neonatal postmortems". *Irene Scheimberg,* UK
16:00-16:30	"When a fetus does not move: fetal akinesia". *Phil Cox, UK*
16:30-18:00	**PPS Annual General Meeting**
21:00	**PPS Annual Dinner at "Casino de Madrid"**

Saturday 27th

09:00-10:30 **Slide Seminar "Unexpected causes of perinatal death"**
Chairpersons: *Christina Vogt* and *Marta Cohen*

- 09:00-09:18 *Peter Nikkels*, The Netherlands
- 09:18-09:36 *Peter Nikkels* , The Netherlands
- 09:36-09:54 *George Kokai*, UK
- 09:54-10:12 *Christina Vogt*, Norway
- 10:12-10:30 *Marta Cohen*, UK

10:30-11:00 **Coffee Break**

11:00-11:45 **Oral Presentations**
Chairpersons: *Phil Cox* and *Irene Scheimberg*

- 11:00-11:15 **O11** - Congenital anomalies and chromosomal abnormalities in fetuses under 20/40 weeks of gestation: An analysis of 482 fetal autopsy cases.
  Tamas Marton, Ann Paraiso, Beata Hargitai, Phillip Cox
- 11:15-11:30 **O12** - Ventriculomegaly with an unusual pattern of intracranial calcification. 3 cases of a possible new syndrome.
  P.M.Cox, I.U.Niklaus-Wollenteit, T.G. Marton
- 11:30-11:45 **O13** - Chronic villitis of infectious versus unknown etiology in autopsy placentas.
  A. Konstantinidou, M. Zafirelli, N. Spanakis, N. Papantoniou, A. Tsakris

11:45-12:00 **Closure and final remarks**
Christina Vogt and *Ronald de Krijger*

12:00-13:30 **IPPA Graduates Slide Seminar**
Chairpersons: *Jean Keeling* and *George Kokai*

15:30-22:00 **Optional Tour to Toledo**

Oral Presentations

O01 - Synchronous pancreatic and colonic adenocarcinoma in a 16 y/o Inuit male with homozygous PMS2 DNA mismatch repair gene deficiency syndrome

Camelia Stefanovici[1], Kris Milbrandt[2], Andrew McKay[2], Michael Cantor[3], Bernie Chodiker[4], Rochelle Yanofsky[5], Albert E. Chudley[4]

Departments of [1]Pathology, [2]Surgery, [3]Gastroenterology, [4]Paediatrics and Child Health and Program in Genetics and Metabolism, University of Manitoba, Winnipeg Manitoba Canada; [5]CancerCare Manitoba, Winnipeg Manitoba Canada

Introduction: PMS2 DNA mismatch repair gene had rarely been considered as a cancer susceptibility locus. Evidence has been gathered?recently that PMS2 mutation, in a homozygous state, leads to a constitutional mismatch repair-deficiency syndrome causing paediatric malignancy. Case report: A 16 y/o adolescent male from Sanikiluaq, Nunavut came to the medical attention because of jaundice and abdominal pain. The patient had several café-au-lait spots. A 5 cm mass in the head of the pancreas was discovered and the fine needle aspiration of the mass showed atypical cells arranged in an acinar pattern, consistent with adenocarcinoma. He underwent a Whipple procedure. This mass was consistent with an ampullary adenocarcinoma. Immunohistochemistry for mismatch repair genes showed intact staining for MLH-1, MSH-1 and MSH6, and absence of PMS2 in both tumor and normal tissue. DNA analysis showed a homozygous splicing mutation in PMS2: 2002 A>G at the end of exon 11, a known founder mutation in this population. A mass in the ascending colon was identified by colonoscopy, confirmed as a pT3 N2a poorly differentiated signet ring cell carcinoma in the total colectomy specimen. The mismatch repair gene profiles were identical to the previous tumor. Discussion: The PMS2 gene encodes the mismatch repair endonuclease PMS2. The PMS2 gene family members are found in clusters on chromosome 7. The protein forms a heterodimer with MLH1 and this complex interacts with MSH2 bound to mismatched bases. As illustrated by this case, defects in this gene are associated with childhood malignancy.

Abstract Sponsor: Camelia Stefanovici

O02 - Study of bax expression by RT-PCR from paraffin embedded pediatric astrocytomas correlates with lower survival in low grade.

[1] Pedro Valencia-Mayoral, [1]Pilar Eguía-Aguilar, [1]Mario Pérezpeña-Díazconti, [1]Fernando Chico-Ponce de León, [2]Miguel Ángel Méndez-García, [1]Francisco Arenas-Huertero.

[1]Hospital Infantil de México Federico Gómez. [2]Universidad Iberoamericana. Mexico City. México

Background: Among the parameters related to recurrence in astrocytomas, the rates of cell proliferation and cell death are cellular responses that play a key role in maintaining integrity in the central nervous system, and the response to therapy. It has been shown that Bax and Bak has significant effects on astrocytic cells in adults, v.g. absence or low expression of Bax is associated with unfavorable prognoses. There are as yet no studies in pediatric patients that suggest a molecular marker related to cell death and has prognostic value. METHODS: Total RNAm was extracted from 54 pediatric astrocytomas, embedded in paraffin and Bax gene was then amplified by RT-PCR. Lately correlations were made between Bax and clinic-pathological data. RESULTS: mRNA was optimally extracted in all samples. Cases positive for Bax survived only half as long as those that were negative for this gene ($p=0.05$), and expression of Bax was associated with recurrent, low grade tumors ($p=0.0173$). Kaplan-Meier survival curves also showed reduced survival times in patients who were positive for Bax ($p=0.037$). A hierarchical regression analysis indicates that Bax is an important predictor of survival ($p<0.05$). CONCLUSIONS: The expression of the RNAm of Bax can be a new marker of poor prognoses in pediatric patients with low grade astrocytomas.

Abstract Sponsor: Mario Perezpeña-Diazconti

O03 - Vascular abnormalities in intestinal atresia

Valencia-Mayoral Pedro, Ramón-García Guillermo, Perezpeña-Daizconti Mario, Haro-Sánchez Victor
Departamento de Patología. Hospital Infantil de México Federico Gómez

Background: Intestinal atresia is a common malformation of small intestine and is the most frequent cause of obstruction in neonates. Several causes have been described having all in common a vascular accident as a plausible explanation for its occurrence. Objective: Description of vascular abnormalities in atretic segments and possible relationship to ischemic and vascular insult. Methods: a descriptive retrospective study. Cases with diagnosis of intestinal atresia were selected from the files of the department of Pathology of the Hospital Infantil de Mexico during a five year period from January 2004 to December 2008. Histological slides were reviewed and we described the vascular changes and correlated with the type of atresia. Results: 20 cases were retrieved. Vascular alterations were found in all them. Angiomatoid formations, irregularities in their walls, congestive dilated vessels, linfangiectasias and smooth muscle and fibroblastic proliferation were found. The types of atresia were, 8 cases type II, 6 cases type IIIa, 3 cases IIIb and 3 cases type IV. Conclusion: Vascular abnormalities are frequent in resected segments of intestinal atresia. They are concordant with a vascular derangement as a pathogenic actor supporting the theory of an ischemic event for this malformation.

Abstract Sponsor: Mario Perezpeña-Diazconti

O04 - Genetic analysis of Pleuropulmonary Blastoma (PPB) by CGH and FISH

Vokuhl C (1), de Leon L (1), Kirsch S (2), Koscielniak E (2), Leuschner I (1)
(1) Department of Pediatric Pathology, University of Kiel, Germany (2) Olgahospital, Pediatrics 5, Stuttgart, Germany.

PPB is a rare malignant intrathoracic tumour primarily affecting children under five years of age. PPB are histologically divided into 3 subtypes: Type I PPBs are composed of cysts, type III is a solid lesion with a variegated morphologic appearence. Type II has a mixed morphology consisting of cystic and solid areas. The genetics of PPB are poorly understood. There are a limited number of karyotype- and CGH-analyses published, showing complex chromosomal changes. We analysed 16 PPB cases of the Kiel Paediatric Tumour Registry by CGH and confirmed some changes by FISH. Frequent findings by CGH were losses on 4q, 5q, 9p and gains on chromosome 8, 17 and 20q. Genomic amplification was observed in 5 cases, four related to 15q25qter, and one to 1p. FISH could confirm seven gains of chromosome 8 (7/16, 44%) and four amplifications of the IGF1R-gene on 15q26 (4/16, 25%). All of the tumours with IGF1R amplification were type III PPBs. One of the PPBs with gain of chromosome 8 was a type II tumour and six tumours were type III PPBs. In our series of 16 PPBs 25% of PPB have an amplification of the IGF1R gene and 44 % show a gain of chromsome 8. All of the tumours with IGF1R amplification were PPB type III, indicating that it is a later event in tumor progression, while the gain of chromosome 8 was found in both type II and type III tumours indicating that these changes are probably earlier events in tumour developement.

Abstract Sponsor: Ivo Leuschner

O05 - Upregulation of HOXB4 plays a role in the pathogenesis of nephroblastomas and is mediated by miR-23a.

Karin Koller[1], Suman Das[1], Ivo Leuschner[2], Melanie Korbelius[1], Gerald Hoefler[1], Barbara Guertl[1]

[1] Institute of Pathology, Medical University of Graz, Auenbruggerplatz 25, 8036 Graz, Austria [2] Kiel Paediatric Tumor Registry, Department of Paediatric Pathology, University of Kiel, Arnold-Heller-Str. 3, 24105 Kiel, Germany

Aim of the study: Nephroblastomas resemble morphologically and genetically different stages of developing kidney. The homeobox family of transcription factors has been shown to play an important role in the development of the kidney. Accordingly, some of the homeobox transcription factors were implicated in the development of nephroblastomas by a microarray analysis. We therefore investigated the role of HOXB4 and its regulation in the pathogenesis of nephroblastomas. Material and Methods: Expression of HOXB4 was investigated by quantitative REALtime PCR and immunohistochemistry in formalin-fixed, paraffin-embedded samples of nephroblastomas. In an in-silico analysis we identified the regulation of HOXB4 by miR-23a. Expression levels of miR-23a were examined by quantitative REALtime PCR. Mechanism of action was verified by a Luciferase-assay and Westernblot analysis. Results: In our samples investigated so far, we identified an overexpression of HOXB4 mRNA in 5 of 34 samples. 14 of 27 nephroblastomas showed a stronger protein expression in comparison to mature kidney. All 21 nephroblastomas investigated had a significantly lower expression of miR-23a in comparison to renal parenchyma. In a cell culture model Luciferase activity was significantly downregulated by miR-23a compared to the scrambled control. Western blot analysis showed a significant downregulation of HOXB4 protein levels by miR-23a accordingly. Conclusion: Our results demonstrate that HOXB4 might play a role in the development of a subset of nephroblastomas. We also demonstrated for the first time the regulation of HOXB4 by miR-23a.

Abstract Sponsor: Barbara Guertl

O06 - Two cases of Leiner's Disease

Helen Wainwright, Roc Kashula
NHLS D7 Anatomical Pathology Laboratory, Groote Schuur Hospital & the UCT Faculty of Health Sciences, Observatory, Cape Town, South Africa. Anatomical Pathology, UCT faculty of Health Science, Cape Town, South Africa.

Leiner disease was first described in 1908 by C. Leiner. The infants were breast fed, had generalized erythroderma, diarrhoea, failure to thrive, recurrent local & systemic infection, marked wasting and died of infection. In 1970 it was shown that there was an abnormality of phagocytosis, some cases were familial and fresh serum normalized phagocytosis. There is a deficiency in complement component C5D. The gene is situated at 9q32.2 which is the same site as Complement C5.
Method: The neonatal and fetal postmortem archives of the Division of Anatomical Pathology were reviewed and 2 cases highly suggestive of Leiner disease were found over a 21 year period out of 3676 postmortems. Results : The first case had parental consanguinity and a family history. The diagnosis was made premortem. No family history was obtained in the second case & a presumptive diagnosis was made at postmortem. Both had generalized skin lesions. The first child died at 2 months, had recurrent infections and died of bacterial bronchopneumonia, and disseminated Candidiasis. The second case was breast fed, had protein losing enteropathy & generalized oedema. She died aged 38 days of bacterial bronchopneumonia & meningitis. Gram positive cocci were present in the skin lesions. Conclusion : The major features suggesting a diagnosis of deficiency of Complement C5 are generalized dermatitis, intractable diarrhoea, recurrent infections, and marked wasting . Molecular diagnosis can now be made from a blood spot. Diagnosis during life is important as treatment is available.

Abstract Sponsor: Helen Wainwright

O07 - Prevalence of duodenal bulb involvement in Coeliac Disease

Sonja Mansfield-Smith(1); Nagendra Rao (2), Mike Thomson (2), Marta C Cohen
(1) Histopathology (1) and Paediatrics (2) Departments. Sheffield Children's NHS FT. Sheffield. UK.

Aim: Identify the value of duodenal bulb (D1) sampling in the diagnosis of CD Vs the more distal duodenum (D2/D3/D4). Material and methods: All biopsies with diagnosis of CD between 2007-2012 with sample of D1 in addition to D2/D3/D4 were retrieved from our files. Cases were classified according to the modified Marsh-Oberhuber classification. Clinical information was retrieved from the notes. Results: 77 children with abnormal modified Marsh-Oberhuber classification grade in whom D1 and D2/3/4 had been biopsied were identified. 17 cases were removed from analysis as the Marsh Grades were 1/2. In 39/ 60c (65%) features were more severe in D1 than D2/3/4 (G1). In 12/39, features of CD were only present in D1. In 8/60c (13%) changes were more severe in D2/3/4 compared with D1 (G2). In 6/8, D1 was normal (Marsh 0) or had non-specific features (Marsh 1). In 13/60c (22%), severity was similar in D1 and D2/3/4 biopsies (G3). A negative serology was identified in 3c of G1, 5c of G2 and 1c of G3; 10c of G1 had weak levels of TTG antibodies. A normal endoscopy was described in 9c in G1, 4c in G2 and none in G3. Diabetes was more frequent in G1 (4c) than in G2 (1c) or G3 (none). Conclusion: The fact that 65% cases had more severe features of CD in D1 and that in 20% of cases these were only present in D1 highlights the relevance of this biopsy site. These patients not infrequently had normal endoscopy and/or negative/weak serology.

Abstract Sponsor: Marta C Cohen

O08 - Type 2 Mosaicism of Nevoid Basal Cell Carcinoma Syndrome

Isabel Colmenero, Fernando Casco*, Angela Hernández-Martín**, Antonio Torrelo**, Luis Requena****

Departments of Pathology* and Dermatology**, Hospital Niño Jesús, Madrid, Spain
Department of Dermatology***, Fundación Jiménez-Díaz, Madrid, Spain

Introduction: The nevoid basal cell carcinoma syndrome (NBCCS) or Gorlin syndrome (GS) is an autosomal dominant disorder resulting from heterozygous mutations in any of PTCH1, PTCH2, SUFU or other yet unknown genes. Its main features include basal cell carcinomas, odontogenic keratocysts, skeletal anomalies, ectopic calcifications and palmo-plantar pits. Some cases of segmentally arranged lesions of NBCCS have been described in the form of type 1 mosaicism. However, so far, no cases of type 2 mosaicism of NBCCS have been recognized. Case: A 12-year-old girl with family history of NBCCS had an odontogenic keratocyst as well as unilateral, segmentally arranged basaloid skin lesions and a blaschkolinear arrangement of palmo-plantar pits. Both the patient and her father carried a heterozygous, single-base substitution mutation in exon 18 of the PTCH1 gene. In the patient's affected skin, a short deletion in exon 3 of the PTCH1 gene was detected; this mutation was ruled out in normal skin and in germline, thus confirming its mosaic state. Discussion: We report herein a patient with family history of NBCCS, with an inherited germline mutation in PTCH1 gene; who had severe, segmentally arranged manifestations of NBCCS in which a second mutation in PTCH1 in a mosaic state was present. A type 2 mosaicism has been demonstrated at a molecular level for the first time in NBCCS.

Abstract Sponsor: Isabel Colmenero, Fernando Casco

O09 - CLOVES and Klippel-Trenaunay Syndromes Share Somatic Mosaic Activating Mutations in PIK3CA

Valerie L. Luks (1,6), Ahmad I. Alomari (2,6), Steven J. Fishman (3,6) Samantha A. Spencer (4,6), John B. Mulliken (5,6), Harry P.W. Kozakewich (1,6), Matthew L. Warman (4,6), and Kyle C. Kurek (1,6).

Departments of Pathology (1), Vascular and Interventional Radiology (2), Surgery (3), Orthopaedic Surgery (4), Plastic Surgery (5), and Vascular Anomalies Center (6), Boston Children's Hospital and Harvard Medical School, MA 02115 USA

Aim: Congenital Lipomatous Overgrowth with Vascular, Epidermal, and Skeletal anomalies (CLOVES) is a sporadically occurring, non-hereditary rare disorder characterized by asymmetric somatic hypertrophy and anomalies in multiple organs. We recently employed massively parallel sequencing of fresh and fixed archival tissue to identify somatic mosaic activating mutations in PIK3CA in individuals with CLOVES syndrome. Klippel-Trenaunay syndrome (KTS) exhibits similar features with CLOVES, i.e. vascular anomalies with overgrowth, but with a more limited distribution of involvement, suggesting KTS involves a common genetic pathway. Materials and Methods: We assessed for 3 common PIK3CA mutations (E542K, E545K, H1047R) in genomic DNA from archival lesional tissue of 10 KTS individuals using competitive allele-specific PCR or PCR amplification followed by subcloning and Sanger sequence analysis. Results: We identified somatic mosaic PIK3CA mutations in lesional tissue from 8 KTS individuals. When assessed, mutant allele frequencies were < 25% in affected tissue from multiple embryonic lineages, similar to findings in CLOVES tissue. Conclusions: CLOVES and KTS are caused by post-zygotic activating mutations in PIK3CA. We are currently performing massively parallel PIK3CA sequence analysis in lesional tissue from additional CLOVES and KTS individuals to determine if genotype-phenotype correlations exist, although the syndromes may also represent an allelic spectrum determined by timing and distribution of mutant cells during development.

Abstract Sponsor: Kyle Kurek

O10 - Splenic cysts associated with hematuria

G. Guarda-Muratori[1], C. Otal-Salaverri[1], A. García-Escudero[1], N. Conde-Cuevas[2], J. Armas-Padrón[1], I. Ortega-Medina[1], P. Gallinato-Perez[1],R. González-Campora[1]
[1]Servicio de Anatomía Patológica. [2]Servicio de Pediatría, Sección de Onco-Hematologia pediátrica. Hospital Universitario Virgen Macarena, Sevilla, España.

Aim of study: report of a case of splenic cysts associated with hematuria. Materials and Methods: Clinical findings: a 10 year-old patient with a sore throat and fever that presented gross hematuria three days later. Physical examination: nontender splenomegaly without hepatomegaly. Laboratory, Serology and X-rays: normal. Urine: glomerular hematuria. No infection; normal glomerular filtration; no proteinuria. Abdominal Ultrasonography: enlarged spleen (16 cm) with two cystic lesions, of 10 cm and 4.3 cm. Abdominal MRI: splenic cysts with hemorrhagic content. Results: Gross features: splenectomy specimen weighing 435 g and measuring 18x12x7cm, that presents two cystic lesions, the largest of 12 cm in maximal diameter. Both lesions have a whitish and smooth internal surface, showing a hemorrhagic content. Microscopic features: Spleen: the cysts are lined by stratified squamous or cuboidal epithelium, with fibrin-hematic content. Granulomatous inflammatory reaction to cholesterol crystals. No parasitic structures. Renal biopsy: mesangial widening involving matrix and cells. Red blood cell casts and few hyaline casts in the tubules. Mild interstitial inflammatory reaction, of lymphocytic type. Blood vessels without significant histological abnormalities. Immunofluorescence: mesangial deposits and capillary loop subendothelial deposits. IgA (+++), Fb (++), IgG (+). Diagnoses: Spleen: non-parasitic epithelial cysts with fibrin-hematic content associated with granulomatous inflammatory reaction (cholesterol crystals). Kidney: mesangial proliferative glomerulonephritis with deposits of IgA (Berger`s disease). Conclusion: The main differential diagnosis must be made with peliosis associated with IgA nephropathy. In our case there was epithelial lining, a histological finding that allows us to rule out splenic peliosis.

Abstract Sponsor: Fernando Casco

O11 - Congenital anomalies and chromosomal abnormalities in fetuses under 20/40 weeks of gestation: An analysis of 482 fetal autopsy cases.

Tamas Marton, Ann Paraiso, Beata Hargitai, Phillip Cox
West Midlands Perinatal Pathology Birmingham Women's Hospital (BWH) Birmingham, UK

Aim A retrospective study of 482 fetal autopsies to establish what proportion of cases have major or minor developmental, or chromosomal abnormalities. Material 476 consecutive autopsies of intrauterine deaths (IUD, 305/476) or miscarriages (171/476), =20 weeks gestation from our database were analysed. Karyotyping was carried out with MLPA/QF-PCR method (PM sample), conventional karyotyping (antepartum sample) and in selected cases CGH microarray studies were done. Results IUD-s: 141/305 (46.2%) had cytogenetic tests, with 25.5% abnormal, 72.3% normal result and 2.2% failed. Major anomalies: 109/305 (35.7%), 91/106 (83%) had a cytogenetic test, with 33/91 (36%) abnormal result, 1 failed. Minor anomalies: 90/305 (29.5%), 33 cytogenetic tests showed 31/33 (94%) normal result and 2 failed. No anomalies: 106/305 (34.8%), 17 cytogenetic tests showed 14/17 (82%) normal, 3 (18%) abnormal result. Miscarriages: 34/171 (20%) had cytogenetic test, with 88% normal result, 9% abnormal result, 1 failed. Major anomalies: 25/171 (14.6%), 12/25 (48%) had cytogenetic test, with 9/12 (75%) normal, 3/12 (25%) abnormal result. Minor anomalies: 62/171 (36.3%), 15/62 (24%) had cytogenetic test, all normal. No anomalies: 84/171 (49.1%), 7/84 (8%) genetic tests, 6 normal and 1 failed. Conclusion IUD was associated with a significantly greater proportion of major congenital anomalies compared with miscarriages (35.7 vs. 14.6%). Only 4 samples have failed out of the 175 cytogenetic samples that confirms MLPA/QF-PCR is a much more efficient method than conventional karyotyping. These data suggest that cytogenetic testing from miscarriages =20 weeks with no congenital anomalies or minor dysmorphic features only is not indicated.

Abstract Sponsor: Tamas Marton

O12 - Ventriculomegaly with an unusual pattern of intracranial calcification. 3 cases of a possible new syndrome.

P.M.Cox, I.U.Niklaus-Wollenteit, T.G.Marton West Midlands Perinatal Pathology, Department of Cellular Pathology, Birmingham Women's Hospital, Mindelsohn Way, BIRMINGHAM, B15 2TG, UK

Introduction We present 3 cases of an apparently novel syndrome of cerebral ventriculomegaly, with Dandy-Walker malformation (DWM) and unusual intracranial calcification that has not, we believe been described previously. Case Histories Cases 1 and 2 were the only children of a Bangladeshi first cousin couple. Ventriculomegaly and a DWM or variant was identified on mid-trimester ultrasound scan. Case 3 was the first child of non-consanguineous East European parents. Mild ventriculomegaly and DWM was identified on scan. Case 1 died in utero at 23 weeks gestation. Cases 2 & 3 were terminations at 23 weeks gestation. Post Mortem Findings All cases showed an enlarged cranium due to hydrocephalus. Cases 1 & 2 exhibited mild limb shortening and case 3 had bilateral talipes. DWM was confirmed in cases 1 and 3, but the vermis appeared normal in case 2. All cases showed histologically normal brain development, with an identical pattern of calcification: a band of calcified axons adjacent to the cerebellar dentate and in the internal capsule; globular calcification in the germinal matrix; calcification in the thalamus and hypothalamus; perivascular calcification in the forebrain, cerebellum and brainstem. In all cases the placental villi showed abnormal maturation suggestive of mesenchymal dysplasia. Conclusion Intracranial calcification is associated with various acquired (infection, ischaemia) and inherited (Aicardi-Goutieres, Cockayne, Fowler) disorders. The pattern of calcification may help to predict the underlying cause. We have not identified previous reports showing similar calcification associated with hydrocephalus and DWM and suggest that this represents a novel autosomal recessive syndrome.

Abstract Sponsor: Phillip Cox

O13 - Chronic villitis of infectious versus unknown etiology in autopsy placentas

A. Konstantinidou, Zafirelli, Spanakis, N. Papantoniou, A. Tsakris
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AIMS: This study aimed a) to investigate the infectious versus unknown etiology of chronic villitis in placentas obtained at fetal autopsy and b) to assess the role of immunohistochemical inflammatory markers in the discrimination between chronic villitis of infectious versus unknown etiology. MATERIAL and METHODS: We studied 72 placentas histologically diagnosed with chronic villitis. All placentas were obtained at autopsy in cases of intrauterine fetal death (IUFD), abortion/premature labour or termination of pregnancy. The investigation for a possible infectious etiology was based on PCR testing for viruses in placental and fetal tissues, histological or immunohistochemical findings of specific infections, and clinical information on maternal serology or bacterial cultures. The immunohistochemical typing of the villous inflammatory infiltrates included 6 markers, CD45, CD3, CD4, CD8, UCHLa and CD138. RESULTS: We found evidence of infectious chronic villitis in 77.8% of the examined placentas versus 22.2% of cases termed as villitis of unknown etiology (VUE). Viruses were the most common infectious agents (73.2%) versus bacterial/parasitic agents (26.8%). Adenoviral genome was the more common among viruses, followed by cytomegalovirus, enteroviruses, parvovirus and herpes simplex virus. Immunohistochemical typing of the inflammatory infiltrates failed to reveal any statistically significant differences between infectious villitis and VUE. CONCLUSIONS: Detailed search for infectious agents in autopsy placentas provides evidence that chronic villitis can be more often attributed to histologically nonspecific viral infection.

Abstract Sponsor: Anastasia Konstantinidou

General Information

Congress dates

October 25th to 27th, 2012

Venue

Hotel Me Madrid Reina Victoria
Plaza de Santa Ana, 14
28012 Madrid - Spain

Language

The official language of PPS Meeting is English. No translation to Spanish or other languages will be available.

Acreditations

Badges and delegate documentation can be collected at the Technical Secretariat. Please it is necessary to wear your name badge to enter the scientific sessions and all areas of the meeting.

Technical Secretariat Schedules

Thursday, 25th October	16:00-20:00
Friday, 26th October	08:30-19:00
Saturday, 27th October	08:30-14:00

Invited Speakers and Oral Authors

After getting the name badge, speakers must download their presentation 45 minutes before the beginning of their session.

Certificates

Attendance and abstract certificates will be provided onsite upon request.

Social program

Welcome Cocktail

Thursday, 25th October (included in the registration fee)
Location: ME Madrid Reina Victoria Hotel
Time: 20:30
Badge required

PPS Annual Dinner

Friday, 26th October (included in the registration fee)
Location: Casino de Madrid
Time: 21:00
Badge required

Optional Tour for Attendees

Toledo Tour
Saturday, 27th October
Meeting point: Main Hall Me Madrid Reina Victoria Hotel
Time: from 15:30 to 22:00
Price: Tickets cost 90 € per person. The tour price is for a minimum of 20 pax. or the cost may vary

Tour for Accompanying persons (Included)

Madrid Panoramic Tour
Friday, 26th October
Meeting point: Main Hall Me Madrid Reina Victoria Hotel
Time: from 09:00 to 13:00

Author list

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Technical Secretariat

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