Abstracts

MYCN gene which was confirmed by FISH. Tumor MYCN amplification status is unknown.

Conclusion Management of perinatal neuroblastoma includes close observation with therapeutic intervention reserved for advanced stage and/or clinical progression. Two of the five cases of neuroblastomas described in patients with dysmorphic features and a germline partial 2p duplication, were detected perinatally. All patients had an aggressive clinical course. In the subset of patients with perinatal neuroblastoma and multiple congenital anomalies, FISH or ACGH testing for partial 2p gain may identify those who may need more aggressive management.

556  IGFR1 DELETION IN A PRENATAL CASE

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Small fetuses constitute a large group including healthy small fetuses, fetuses suffering from utero-placental insufficiency or chromosomal abnormality. In the latter case, chromosomal rearrangements may be showed by fetal karyotyping. However, they are usually cryptic and require to be detected by molecular investigations.

In this study, we report on a case of 15q26 microdeletion diagnosed prenatally in a fetus found to have severe intra uterine growth retardation, congenital diaphragmatic hernia and polycystic kidneys identified at 28 weeks of gestation by ultrasonography.

Ammiocentesis was performed and revealed a normal karyotype of 46, XX. However, phenotypic features let us to test the 15q26 microdeletion which was confirmed by IGFR1 FISH probe.

IGFR1 gene is involved in pre and post natal growth. Monosomy for IGFR1 gene is responsible for growth delay, microcephaly, mental retardation, micrognatha and deafness. Congenital diaphragmatic hernia and polycystic kidneys are common findings in 15q26 microdeletion. Genotype phenotype correlation localized critical region implicated in diaphragmatic hernia in 15q26.1–q26.2. NR2F2, CHD2, RGRMA and SIAT8B are considered to be most likely candidate genes. Genes involved in kidney defect are little studied. In our case, the deletion must be extended to 15q26.1. Molecular characterization by CGH array is considered to refine genotype phenotype correlation and to localize candidate genes for diaphragmatic hernia and kidney defect.

15q26 microdeletion causes intra uterine and post natal growth retardation variably associated to other malformations. We highlight the importance of molecular analysis in the prenatal diagnosis of cryptic chromosomal abnormalities.

557  SUPPOSED"SATELLITE 8q" CHROMOSOME ASSOCIATED TO A GROWTH RETARDATION AND NEUROLOGY DEVELOPMENT DELAY

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Polymorphisms are cytogenetic laboratory findings that in most cases are inherited without clinical repercussion, when they are found in a karyotype usually none additional studies are carried out. One of the most common is the presence of a satellite (NOR: nucleolus organizer regions) on a non-acrocentric chromosome. These extra NOR regions generally result from a translocation between a NOR region of an acrocentric chromosome and a non-acrocentric chromosome and are easily confirmed by NOR-bands in laboratory. These translocations are usually terminal and have been described on multiple chromosomes, the most frequent involves de Y chromosome. We present a case with a satellite chromosome and clinical expression. A 24 months old girl was referred to our hospital with deeply postnatal growth retardation, dysmorphic features, motor delay and seizures. Cytogenetic studies were requested and a satellite in a chromosome 8 (8q2) was founded. FISH showed on the abnormal chromosome 8 a hybridization signal much bigger than the one detected on the normal homologue, these suggest a 8q24 inverted duplication. Array-CGH analysis at 40 Kb resolution confirmed the duplicated region of 17.2 Mb in 8q24.1–q24.3, at a terminal to 55 Kb deletion. Furthermore, a triplicated region of 76 Kb between the duplicated and deleted regions was detected. The diagnosis is a 46, XX inv dupdel (8) de novo. With this case we want to show that properly cytogenetic studies are very important in order to define the “supposed satellite chromosome” and to find other chromosome abnormalities that could explain the clinical findings of some patients.

558  PRADER-WILLI SYNDROME DUE TO MATERNAL UNIPARENTAL DISOMY FOLLOWING ASSISTED REPRODUCTIVE TECHNOLOGY

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Assisted Reproductive Technology (ART) refers to methods used to provide pregnancy by the manipulation of sperm and ovum in vitro. Concern has been raised recently on the safety about the health of children born after ART. Although abnormal genomic imprinting has been shown to be involved in a number of genetic syndromes identified in the pregnancies with ART, there is a lack of evidence linking ART with the Prader-Willi syndrome (PWS) which is caused by the lack of expression of paternally inherited genes on chromosome 15q11–q13. Paternal microdeletion (70%), maternal uniparental disomy (25–30%), and imprinting defect (2–5%) are the main causes of PWS. Here 2-year-old male/female twins, male with upd(15)mat, born to a nonconsanguineous parents following ART is presented. The proband was between 90–97 percentile for height, weight and head circumference on admission. Physical examination showed hypotonia, almond-shaped palpebral fissures, low-set ears, long philtrum, small hands and feet. His history revealed feeding difficulties and delayed developmental milestones. MRI, EEG and EMG were normal. Echocardiography showed supravalvular pulmonary stenosis, which disappeared with aging. Pelvicactasis was demonstrated sonographically. Serum biochemical tests, thyroid function tests and amino acid chromatography were normal. Karyotype analysis, FISH analysis for PWS and subtelomeric regions were normal. DNA methylation analysis revealed maternal uniparental disomy. In conclusion this case contributes to the literature in two ways, first the importance of clinical evaluation with molecular testing in the diagnosis of PWS is pointed out, and second ART may have an effect on the occurrence of imprinted diseases such as PWS.

559  WILLIAMS SYNDROME PRESENTING WITH FINDINGS CONSISTENT WITH ALAGILLE SYNDROME

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WILLIAMS SYNDROME PRESENTING WITH FINDINGS CONSISTENT WITH ALAGILLE SYNDROME

WILLIAMS SYNDROME PRESENTING WITH FINDINGS CONSISTENT WITH ALAGILLE SYNDROME
Williams syndrome is a rare genetic neurodevelopmental disorder with a characteristic physical and behavioral phenotype caused by deletion at the 7q11.23. It is usually diagnosed in childhood by clinical evaluation when typical facial features, supravalvular aortic stenosis on echocardiography, hypercalcaemia and other neurodevelopmental and behavioral profile may become apparent. Conjugated hyperbilirubinemia, posterior embryotoxon, pulmonary stenosis, vertebral anomalies, renal anomalies and vascular anomalies are typical features of Alagille syndrome, which is caused by mutations in or deletion of the JAG1 gene at 20p12 or rarely the NOTCH2 gene at 1p12. There may be some overlap in the clinical features between these syndromes; however, conjugated hyperbilirubinemia, posterior embryotoxon and vertebral anomalies are not features of Williams syndrome. The typical facial features specific to each of the syndromes usually become apparent with age and pose a challenge in making a diagnosis in the newborn period and especially when the baby is premature. We report a preterm newborn with spectrum of clinical features highly suggestive of Alagille syndrome but array CGH consistent with Williams syndrome. To the best of our knowledge, this very unusual association, has been reported only on three occasions in the past and further extend the phenotype of Williams syndrome.

**EFFECT OF TRANEXAMIC ACID IN THE MANAGEMENT OF HEMOPHILIA**

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**Background and Aims** Hemophilia is a rare genetic disease. Treatment of hemophilia is a great burden to the patient, family as well as to the nation. Recombinant factor concentrates, currently available products are viral pathogen-free, although there is debate about the risk of transmission of parvovirus B19 and prion pathogens. Because of this very small risk, recombinant factor is the treatment of choice in hemophilia patients.

In developing country like Nepal the treatment is based on the blood product like fresh frozen plasma and cryo precipitates. Recombinant therapy is very expensive and not readily available in local market.

Treatment with tranexamic acid has been tried with success in the management of minor bleedings at hemophilia care unit, Kathmandu Medical college teaching hospital which has reduced the necessity of use of blood product and the cost of treatment.

**Aim** of this study is to see the effect of tranexamic acid in oral hemorrhage, to reduce the cost of treatment to avoid blood product.

**Method** Retrospective study.

**Results** Bleeding stopped in all patient with gum bleeding within few hours of treatment whereas bleeding did not stopped in any patient with tongue injury.

**Conclusions** Extremely useful in the control of mucous membrane bleeding. Main advantage is inexpensive and no risk of bloodborne viral infections.

**For oral presentation**

**Disclosure of financial relationships** There was no financial support from any manufacturer/supplier of the commercial products related to this work.

**PRESENTATION OF RARELY SEEN GASTROINTESTINAL TELANGIECTASIA IN A 4 YEAR OLD CHILD WITH RARE CONDITION GLANZMANN’S THROMBASTHENIA**

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A 4 year old girl diagnosed with Glanzmann’s Thrombasthenia at the age of 5 months had been admitted on previous occasions to hospital with epistaxis.

This encounter describes her presentation with first episode of haematemesis. No focal bleeding source was noted on ENT examination.

Emergency endoscopy showed discrete telangiectasia in stomach. The combination of GI telangiectasia and Glanzmann’s Thrombasthenia has been rarely reported.

**RADIOLOGICAL EVALUATION OF PEDIATRIC CONGENITAL URINARY TRACT ANOMALIES**

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**Purpose** To illustrate a wide spectrum of congenital anomalies of the urinary tract in children.

**Materials and Methods** We demonstrate radiological evaluation and its clinical significance of congenital anomalies of the urinary tract in pediatric patients.

**Results** Demonstrated various pediatric congenital urological anomalies included kidney (renal agenesis, ectopic kidney, multicystic dysplastic kidney, duplication), ureter (primary megaureter, ectopic ureteroceles, ectopic insertion of ureter), bladder (anterior bladder diverticulum), and urethra (posterior urethral valve, urethral diverticulum, urethral polyp). We also described its clinical significance.

**Conclusion** Radiological evaluation including Ultrasonography, CT, and/or MRI is very useful for diagnosis and follow-up of pediatric urologic structural anomalies.

**CASE REPORT: TRANSVERSE MYELITIS CAUSED BY ENTEROVIRUS INFECTION**

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**Background and Aims** Enterovirus infections are most common in young children, and acute infection of the CNS occurs at all ages. Meningitis is by far the most common CNS manifestation. Certain enteroviruses (ie, polioviruses, enterovirus 71) preferentially target the motor nuclei and anterior horn cells of the brain and spinal cord, causing acute paresis of cranial and spinal nerves.

Our objective is report an uncommon case of a teenage girl with transverse myelitis caused by enterovirus infection whose obtained good outcome after plasmapheresis treatment.

**Methods** Report case.

An 11 years old girl with lower limb paresthesia, are flexia, hypotony, evolving with bladder’s urine retention, paresis and respiratory effort about 10 days. The liquor exam was positive for enterovirus PCR (polymerase chain reaction) and thoraco lumbar NMR revealed transverse myelitis involving C8 and L4 level.