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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, hypercalcemia 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 20p.12 or rarely the NOTCH 2 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 William 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.

560 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

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561 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.

562 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 ureterocele, 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.

563 CASE REPORT: TRANSVERSE MYELITIS CAUSED BY ENTEROVIRUS INFECTION

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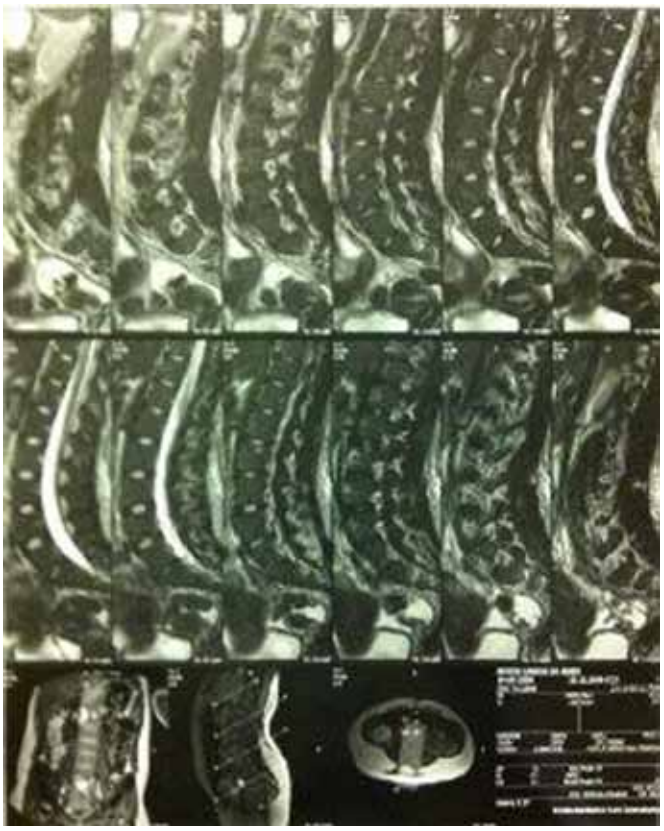
Background and Aims Enterovirus infections are most common in young children, and acute infection of the CNS occurs at all ages. Meningitis is by far the most common CNS manifestation. Certain enteroviruses (ie, polioviruses, enterovirus 71) preferentially target the motor nuclei and anterior horn cells 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 raction) and thoraco - lumbar NMR revelead transverse myelitis involving C3 and L4 level.



Abstract 563 Figure 1 NMR

Results First treatment was initiated with 2 immunoglobulin's cycles and pulse therapy with methylprednisolone not getting satisfactory motor response. The patient developed with respiratory effort and hypoventilation (PCO2 max - 90mmHg) becoming necessary to introduce a course of 5 days of non invasive ventilation. Subsequently, a 5-day plasmapheresis cycle was iniciated, solving the clinical case with an increasing recovering of motor and ventilation function after 1 week of treatment.

Conclusion Enterovirus infections can cause several clinical manifestations such as transverse myelitis. In most of tranverse myelitis cases the treatment is immunoglobulin and pulse therapy with methylprednisolone, yet this case was only solved after plasmapheresis cycle.

564 AUTOIMMUNE ENCEPHALOPATHY NINE YEAR OLD BOY WITH NO HISTORY OF INTEREST

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Background and Aims Nine year old boy admitted with meningeal syndrome. six days ago began with fever being treated with azithromycin. three days before entering a high fever, bilious vomiting, no diarrhea, the next day associated dizziness, headache, drowsiness, the day before admission diplopia and pain associated neck.

Strikes disease, no rashes or petechiae.

Methods Hemogram: 13.24 leukocytes, neutrophils 87.4%

Cerebrospinal fluid red blood cells 160/mmc 30/mmc polynuclear leukocytes 5%, lymphocytes 95%, Gram negative, PCR Herpes simplex I and II negative, negative enterovirus, varicella zoster negative. Negative blood cultures, Mantoux negative.

Abnormal EEG tracing during wakefulness slow waves of high amplitude delta, acute, which are located in anterior and temporal area.

RMN ill-defined hyperintense areas in pons, cerebellum, basal ganglia, right parietal subcortical white matter. suspected encephalitis retrovirus by areas of parenchymal signal alteration above and infratentorial level.

Three days later reduction in the number and extent of intraaxial lesions above and infratentorial.

Results The clinical and resonancia were doing suspect herpes virus encephalopathy, so income at the start of treatment with intravenous acyclovir. When we receive negative results and the improvement of symptoms, treatment it was suspended treatment with acyclovir on the fifteenth day and start treating autoimmune encephalitis with five boluses of methylprednisolone one gram every 24 hours.

Conclusions The day of discharge was treated with 60 mg of prednisolone daily. the fever subsides completely within three days before discharge, and intention tremor persists discrete gait instability.

565 REFLECTION ON A CASE OF DOPAMINE-RESPONSIVE DYSTONIA

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Background Characteristic symptoms of Dopamine-responsive dystonia (DRD) are increased muscle tone and Parkinsonian features. Children with DRD are often misdiagnosed. The disorder responds well to treatment with Levodopa.

Aim To reflect on a case of DRD.

Method Case report of a 13 years old girl misdiagnosed with tetany.

Results The girl was hospitalized for opisthotonus, positive Trousseau and Chvostek signs, diagnosed as tetany. The laboratory analysis have shown: normal serum of calcium (2.28 mmol/l), normal serum of magnesium (0.80 mmol/l), normal serum of phosphor (1.26 mmol/l), normal alkaline phosphatase (261 u/l) and normal PTH (27.9 pg/ml). Although the initial evolution was favorable (with intravenous calcium gluconate), the hypoparathyroidism diagnosis requiring reconsideration. The final diagnosis was DRD with long good evolution after Levodopa treatment. Referring to family history we learned that the patient have a cousin with the same symptoms.

Conclusions The misdiagnosis results from the following similarities: increased muscle tone with opisthotonus, writer's cramp with Trousseau sign, facial dystonia with Chvostek sign and difficult speech (due to facial dystonia) with patient illiteracy. All these similarities delayed the DRD diagnosis.

566 KLIPPEL TREUNANAY SYNDROME IN DIFFERENTIAL DIAGNOSIS OF CEREBRAL PALSY

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In cerebral palsy (CP) atrophy of the paretic body half results in disturbed growth. Disturbed growth is also a feature of a rare disorder; Klippel Treunanay syndrome (KTS). Here we report a child with an initial diagnosis of CP because of limping and thinning of the extremities on the right side who had a final diagnosis of KTS. Five year old male was admitted to our department of Pediatric Neurology. He had been followed up with the diagnosis of CP since he started walking because of limping and thinning of the extremities on the right side of his body. His perinatal and natal period was uneventful. Developmental milestones were normal. On physical examination hypertrophy of the left upper and lower extremities