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, supravalvar 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 20p12 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 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.

**Method**

**Results**

Bleeding stopped in all patient with gum bleeding within 5 days. There was no financial support from any manufacturer/supplier of the commercial products related to this work.

**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.

**561**

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

**doi:10.1136/archdischild-2012-302724.0561**

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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**

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

**doi:10.1136/archdischild-2012-302724.0562**

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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.

**563**

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

**doi:10.1136/archdischild-2012-302724.0563**

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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 racton) and thoraco-lumbar NMR revelead transverse myelitis involving C5 and L4 level.
Methods

Neck.

Drowsiness, the day before admission diplopia and pain associated ing, no diarrhea, the next day associated dizziness, headache, azithromycin. three days before entering a high fever, bilious vomit-
geal syndrome. six days ago began with fever being treated with area.

tive. Negative blood cultures, Mantoux negative.

simplex I and II negative, negative enterovirus, varicella zoster nega-
tions such as transverse myelitis. In most of transverse myelitis cases
ction function after 1 week of treatment.

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

Results First treatment was initiated with 2 immunoglobulin’s
cycles and pulse therapy with methylprednisolone not getting satis-
factory motor response. The patient developed with respiratory
effort and hypoventilation (PCO2 max - 90mmHg) becoming neces-
sary 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 ventila-
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Conclusion Enterovirus infections can cause several clinical manifes-
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Abstracts

Abstract 563 Figure 1  NMR

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the treatment is immunoglobulin and pulse therapy with methylpred-
nisolone, yet this case was only solved after plasmapheresis cycle.

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

doi:10.1136/archdischild-2012-302724.0564

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Background and Aims Nine year old boy admitted with menin-
geal syndrome. six days ago began with fever being treated with azithromycin. three days before entering a high fever, bilious vomit-
ning, 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 polynu-
clear leukocytes 5%, lymphocytes 95%, Gram negative, PCR Herpes simplex I and II negative, negative enterovirus, varicella zoster nega-
tive. 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 intra-
axial 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 pred-
nisolone 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 fea-
tures. 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 Trous-
seau and Chvostek signs, diagnosed as tetany. The laboratory analy-
sis 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 ul) 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 similari-
ties: 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 simi-
narities 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 disor-
der; Klippel T reunanay syndrome (KTS). Here we report a child
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 similari-
ties: 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 simi-
narities delayed the DRD diagnosis.

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