polyneuropathy associated with bilateral striatal necrosis. Using homozygosity mapping the pathogenic homozygous missense mutation c.373G>A was identified. This mutation alters a highly conserved glycine residue at position 125 to serine in the *SLC25A19* gene which encodes the mitochondrial thiamine pyrophosphate transporter. *SLC25A19* mutation was previously reported only in the Amish congenital lethal microcephaly but our patients' phenotype is markedly different with normal head circumference, early childhood development, and age-appropriate cognitive skills. Based on the role of SLC25A19 in mitochondrial thiamine transport, supplementation with high dose thiamine was initiated in all patients. Preliminary results suggest a favorable effect.

Our report suggests an allelic phenotype in *SLC25A19* gene defects. Determination of the *SLC25A19* sequence may be warranted in patients with bilateral striatal necrosis or progressive polyneuropathy.

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A NOVEL DE NOVO MUTATION OF CHROMOSOME 7 [46,XX,DEL(7)(P14.2 P15.1)] IN A CHILD WITH FEEDING PROBLEMS

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Background: The phenotype and severity of symptoms associated with mutations of chromosome 7 are directly proportional to the number of bases away. Distal and interstitial deletions are described in 40 cases.

Aim: To report a child with a novel de novo mutation of chromosome 7 [46,XX,del(7)(p14.2 p15.1)].

Clinical case: Female, born at 38 weeks with RCIU and feeding problems with episodes of cyanosis after feedings and failure to thrive. Physical examination showed low implantation of ears, hypertelorism, oblique palpebral fissures, retrognathia, and palate ogived, insertion anomalies of the toes, poor facial expression and mild axial hypotonia.

A transfontanelar ultrasound, MRI, bronchofibroscopy and metabolic study were normal. She was hospitalized until the 32th day of life.

She starts speech-language therapy and presented improvements in swallowing. The PEG was removed at 36 months.

She had recurrent UTI with normal DMSA but with a vesicoureteral reflux (grade III). She has taken UTI antibiotic prophylaxis.

Imagiological studies reveal a bilateral osteonecrosis of femoral epiphysis (Legg-Calvé-Perthes disease).

Nowadays (4years-old) she is being normally fed (BMI=15.8kg/m^2). She has a mild delay of psychomotor development impairment (GQ: 74.1) and some speech problems.

Conclusion: This is the first case report of a patient with this de novo mutation of chromosome 7. This rare mutation was a cause of severe feeding problems in the first years of life.

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THE INFLUENCE OF DOWN SYNDROME ON DEVELOPMENT OF PRIMARY (DECIDUOUS) TEETH

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Down Syndrome (DS) (Trisomy 21) is a relatively common anomaly occurring in one in every 600-700 live births and characterized by impaired growth and development and early senescence. Signs of abnormal development have been observed on ultrasound as early as the first trimester. By the second trimester additional signs are evident on ultrasound, affecting skeletal growth as well as other organ systems. Dental anomalies include reduced tooth size and number, thin enamel and abnormal crown form as well as taurodont roots and a high frequency of missing teeth. The severity of the dental defects varies along the tooth row with later developing teeth most severely affected. Recent studies carried out on exfoliated (shed) deciduous

teeth using a combination of serial microCT scans and SEM have enabled us to reconstruct the chronology and extent of intrauterine and infant growth insults in DS infants, expressed in growth and differentiation of the developing teeth. Our results for the deciduous teeth show that in DS, there is accelerated dental development in the first trimester followed by progressive growth retardation associated with impaired differentiation of the dental tissues. These changes appear to primarily affect cells derived from ectoderm affecting cell division and differentiation resulting in a smaller modified tooth germ and impaired enamel matrix formation and bio-mineralization. The resulting enamel is thin and hypomineralized and this may contribute to the severe abrasion seen in the teeth of DS children despite the associated lack of muscle tone.

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USE OF EMERGENCY O NEGATIVE BLOOD TRANSFUSION IN PAEDIATRIC INTENSIVE CARE UNIT

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Background: In emergency situations O negative blood (O^-) transfusion may be required before standard compatibility tests can be completed. An increase in the use of emergency O^- in our paediatric intensive care unit (PICU) triggered an audit to evaluate our approach and measure this against any identified guidelines.

Methods: PICU patients issued emergency O^- between January 2004 and December 2008 were identified. Case notes were analysed to ascertain the indications for emergency transfusion, time from PICU admission to transfusion, volume and duration of transfusion and the presence of any transfusion reaction.

Results: Eight patients received a total of 10 units. Two patients were transfused twice. The patients median age was six months. Visible active bleeding with low cardiac output was recorded as an indication on three occasions. Five transfusions occurred in children with low cardiac output and suspected but not proven active bleeding. Two further patients were transfused for low Hb only. Up to 20 ml/kg were given as a bolus over 10 to 20 minutes in all but two cases where it was transfused over 90 and 150 minutes. All patients had been admitted to PICU more than four hours previously. In three instances, crossmatched blood was already available. No transfusion reactions were documented.

Conclusions: Published data is scarce with regards to the use of emergency O^-. Our audit demonstrates the variety and inconsistency of practise. Improved local and national guidance would lead to better understanding and safe transfusion practise in the PICU and wider acute paediatric setting.

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PATTERNS OF MINOR BLEEDING IN NEONATES WITH SEVERE THROMBOCYTOPENIA: A PROSPECTIVE OBSERVATIONAL STUDY

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Background and aim: A prospective cross sectional observational study to determine incidence and pattern of minor bleeding in neonates with severe neonatal thrombocytopenia (SNT) defined as platelet count < 60 $x10^9$/L.

Methods: Neonates admitted to seven UK tertiary neonatal units (March 2005 -November 2006) documented to have SNT were enrolled for daily data collection.

Results: 169 babies with SNT were enrolled for 2055 study days. 123 (73%) babies showed minor bleeding, 15 (9%) minor and major bleeds, 31 (18%) no evidence of bleeding. Bleeding as a percentage of study days occured with greatest frequency in extremely preterm infants: < 28 weeks, 44%; 28-34 weeks, 27%; >34 weeks, 28%. Neonates with early SNT (< 3 days) most commonly had hematuria (31%), oozing (20%) and/or nasogastric bleeding (17%). Neonates with late SNT(≥3 days) most commonly showed hematuria (42%), minor bleeding from the endotracheal tube (24%) and oozing (13%). Minor bleeding (bleeding days as a percentage of study days) with SNT was commoner in the first 10 days (50%) compared to >10 days (33%). Neonates with major bleeds did not show a higher incidence of minor bleeding events prior to the major haemorrhage. 41% of infants with nadir