probe. Average gestational age was 29+6 and mean birth weight was 1296g. Our study is ongoing.

Results: Portal venous gas was detected on abdominal ultrasound in 2 out of 6 of the babies. Both babies were nursed in close proximity in the neonatal unit and developed NEC within 72 hours of each other. A third baby with presumed NEC but no PVG-US presented within the same period in the same 13-bedded unit. This subset may represent crossover infection. One of the confirmed NEC cases died post open laparotomy.

Discussion: NEC remains an enigma in neonatal intensive care units. Abdominal ultrasound to detect portal venous gas is a simple, cost-effective, non-invasive, bedside test that is a useful aide in the diagnosis of suspected NEC. A negative scan may permit us to feed those babies with suspected NEC.

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UPPER GASTROINTESTINAL BLEEDING BETWEEN PREMATURE AND MATURE BABIES IN EARLY INFANCY

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Aims: Upper gastrointestinal (UGI) bleeding is not rare among pediatric patients. However, there are limited reports describing this clinical condition in young infants. This study was aimed to assess and analyze the characteristics of UGI bleeding in infants, and try to define the differences between prematurely and maturely born patients.

Methods: A retrospective review for admission up to the age of 6 months with the clinical diagnosis of UGI bleeding along with positive fetal hemoglobin confirmed by Apt-Downey test in newborns or mucosal lesions seen in the UGI tract by endoscopic examination between June 2000 and October 2007.

Results: Thirty patients were enrolled, 14 premature and 17 mature infants. More than half of the patients got the disease before 2 days of age, with more than two thirds before one month. UGI bleeding was more prevalent in premature boys ($p=0.04$). Most premature infants bled from their UGI tract with concomitant disease, mainly respiratory distress syndrome under airway positive pressure support. Hemorrhagic gastritis was found

in 57% of the cases who received UGI endoscopy, being the most frequent. UGI bleeding stopped in 2 days on average after treatment. No recurrent UGI bleeding occurred in those patients followed for about 3 years.

Conclusions: Reason for UGI bleeding is still obscure in neonates and young infants. Premature boys seem to be more vulnerable. Conservative treatment made UGI bleeding evolve rapidly and favorably in early infancy. Both premature and mature patients recovered from UGI bleeding satisfactorily and showed no significant differences clinically.

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FAMILIAL LIPOPROTEIN LIPASE DEFICIENCY

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Background: Mutations in the LPL gene cause familial lipoprotein lipase deficiency. Symptoms of familial LPL deficiency usually begin in childhood and include abdominal pain, acute and recurrent inflammation of the pancreas, skin lesions called eruptive cutaneous xanthoma and an enlargement of the liver and spleen. The risk for two carrier parents to both pass the defective gene and, therefore, have an affected child is 25% with each pregnancy.

Aims: Early diagnosis, routine surveillance and treatment of familial LPL deficiency may help to manage some of the symptoms and sometimes prevent related problems.

Methods: Clinical genetic testing for familial lipoprotein lipase deficiency may be available through an in person genetic consultation for children who are considered at risk. Triglycerides and total cholesterol were measured using commercially available kits (Boehringer Mannheim).

Results: Episodes of abdominal pain are common. Intensity, duration, and localization of episodes are variable. Enlargement of the liver and spleen occurs particularly among affected infants and children. The enlargement of these organs may vary, often in parallel with the fat content of the diet. The risk is the same for boys and girls.

Conclusions: Familial lipoprotein lipase deficiency is an inherited condition that disrupts the normal breakdown of fats in the body. It is characterized by absence of lipoprotein lipase activity and a massive accumulation of chylomicrons in plasma and a corresponding increase of plasma triglyceride concentration. Higher levels of plasma LPL activity are associated with decreased TG and increased HDL cholesterol levels in children.

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RARE GENETIC ANOMALIES IN A CASE WITH GROWTH RETARDATION

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Background and aims: To emphasize the genetic background of a child with growth failure.

Methods: The authors have evaluated a 7 year-old boy in context of failure to thrive. The family history: no related parents. The clinical exam: normal proportioned body, height -3.73 SD, weight -2.078 SD, microcephaly, face dimorphism (down-slanted palpebral fissures), 2nd and 5th fingers clinodactily for both hands, mental retardation, hypoprosopia and aggressive behavior. Achondroplasia and familial short stature were excluded based on clinical criteria.

Results: The blood tests revealed negative serology for celiac disease. The sweat test was normal. Endocrinological evaluation: normal thyroid gland function, low levels for growth hormone and IGF-1 (baseline and after clonidine and insulin stimulation tests). Regarding imagistic investigations: delayed skeletal maturation and cerebral IRM without brain or sella anomalies. Short stature associated with mental disabilities have justified genetic tests (karyotype) identifying double translocation between 1 and 13 chromosomes and between 5 and 11 chromosomes [46,XY, t(1:13) (p31.2; q32), t(5;11) (q14;q25)]. Due to normal parents' karyotypes

we consider this genetic anomalies as "de novo". The growth hormone therapy was followed by improvement of growth velocity.

Conclusions: The authors present a child with short stature secondary to growth hormone deficiency associated with very rare and "de novo" genetic anomalies. There is a chance that one of the four breakpoints disrupts a major gene on 1p31.2 or 13q32 or 5q14 or 11q25. Another possibility is a submicroscopic deletion on one of these four loci, justifying further cytogenetic investigations.

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NEWBORN FEMALE INFANT WITH HAEMOPHILIA A PHENOTYPE ASSOCIATED WITH NOVEL GENOTYPE

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Background: The differential diagnoses for reduced Factor VIII activity/level in a female infant include Turner's syndrome and Normandy (type 2N) variant of Von Willebrand's Disease (VWD).

Case: We present a female infant with low factor VIII activity/levels and raised APPT. She presents in a pedigree comprising both Haemophilia A and VWD.

Genetics: CGH array confirmed XX constitution and DNA testing excluded Normandy variant.

Mutation screening of the F8 gene showed the infant to be heterozygous for a c.2128G>A substitution. This novel base change predicts the replacement of the native Glycine residue at codon 710 with an Arginine residue; p.Gly710Arg. Although this variant has not previously been described, in-silico analysis supports its potential for pathogenicity as Haemophilia A. Two additional variants (c.3780C>G and c.1010-27G>A) were detected on the alternate allele. These polymorphic variants occur in linkage, and have been reported to be associated with reduced Factor VIII levels. Atypical Lyonisation was confirmed by X-inactivation analysis of peripheral blood.