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

S.I. Iurian^{1,2}, V. Belengeanu^{3,4}, M.B. Neamtu^{1,2}, V. Bunesu², B.I. Mehedintu^{1,2}

¹Lucian Blaga University, ²Pediatric Clinic, Sibiu, ³University of Medicine, ⁴Genetic Department, Timisoara, Romania

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

B. Jani¹, S. Kamal-Uddin², E. Gillian³, J. Cutler⁴

¹Oliver Fisher Neonatal Unit, Medway Maritime Hospital, Gillingham, ²Department of Paediatrics, NHS Royal London Hospital, London, ³Kent Haemophilia Centre, Kent and Canterbury Hospital, Canterbury, ⁴Haemophilia Reference Centre, St Thomas' Hospital, London, UK

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.