performance on the Multi-Luminance Mobility Test (MLMT) at 7 standard light levels as measured by a change in score. Additional endpoints were full-field light sensitivity threshold (FST) testing, visual acuity (VA), and Goldmann kinetic VF (GVF), each averaged over both eyes. Safety outcomes included adverse event reporting, laboratory testing, and changes in physical and ophthalmic examinations.

Results For OI patients at Year 5 (n=18) and DI patients at Year 4 (n=8), the mean (standard deviation) MLMT bilateral light level score change was 1.6 (1.1) and 2.4 (1.5) levels, respectively, compared with baseline. Subsequent to the 1-year outcomes, a change of 1 light level occurred in 6 patients (none were below pre-treatment performance) and no change in the remaining 20 (N=26). Mean change in white light FST in log10 (cd.s/m²) averaged over both eyes was -2.02 (1.45) log10 at Year 5 for OI patients (n=17) and -2.58 (1.04) log10 at Year 4 for DI patients (n=8). Mean change in VA (Holladay Scale) averaged over both eyes (logMAR was -0.00 (0.64) at Year 5 for OI patients (n=18) and -0.06 (0.26) at Year 4 for DI patients (n=8). Mean change in GVF III4e sum total degrees averaged over both eyes was 166.6 (208.7) at Year 5 for OI patients (n=15) and 178.8 (241.9) at Year 4 for DI patients (n=8). Five years after treatment, the safety profile (N=29) was consistent with vitrectomy and subretinal injection procedure with 2 reports of cataract, 1 of ptosis, and 1 new report of retinal detachment since the last update. No deleterious immune responses were reported.

Conclusion Improvements in ambulatory navigation, light sensitivity, and VF are maintained for at least 5 years after VN administration in most OI patients.

Improvements in DI patients were consistent with those observed in OI patients. The safety profile of VN is consistent with the administration procedure.

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THE FIRST CLINICAL CASE OF RARE FORM OF FOCAL EPILEPSY CAUSED BY THE NOVEL MUTATION IN THE NPRL3 GENE IN RUSSIAN FEDERATION AND KAZAKHSTAN

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Objective Mutations in the NPRL3 gene (OMIM 600928) are described predominantly in patients with autosomal dominant focal epilepsy according to the HGMD database.

Methods We want to introduce the clinical case of the female patient, 2 years old, with focal epilepsy from healthy parents. She was born full-term from the 1st pregnancy by caesarian delivery. The cerebellum hypoplasia was suspected during ultrasound and MRI diagnostic at 17th week of gestation.

Weight at birth - 3.780 kg, 7/8 on Apgar scale. Neurosonography at birth has shown left-sided ventriculomegaly. First recurrent afebrile tonic extensor epileptic spasms with head and eyes turns to the left side were noted at the age of 2 months. Eyelid and tongue myoclonias were revealed as well. Later the frequency of seizures has increased to 150 per day. The video electroencephalogram (VEEG) showed sharp, spike waves with partial origin at left temporal region, while clonic seizures in the left arm were recorded. The brain MRI has shown focal cortical dysplasia in the left insular region. Signs

of intellectual impairment and behavioural disturbances were revealed during the neurological examination at the age of

24 months. The patient could not walk without support. We have performed full exome sequencing for our patient to identify the molecular genetic causes of the disease after medical genetic counseling.

Results The nucleotide variant c.481C>T which leads to stop codon p.Q161* in heterozygous state was revealed in exon 5 of the NPRL3 gene. This variant was not described in gnomAD and HGMD databases and was considered as pathogenic according to ACMG criteria. An interesting fact is that the most frequent pathogenic variants in the NPRL3 gene (among the 21 described variants in the HGMD) are nonsense mutations and frameshift deletions.

Conclusion This paper describes the first clinical case of rare form of focal epilepsy caused by the novel mutation in the NPRL3 gene in Russian Federation and Kazakhstan.

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FABRY'S DISEASE WITH MINIMAL MANIFESTATIONS IN GIRLS (CLINICAL CASES)

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Fabry disease (Anderson-Fabry disease) is a rare inherited lysosomal multisystem disease with X-linked inheritance due to the deficiency of lysosomal enzyme α -galactosidase A and leads to accumulation of sphingolipids (globoside, globotriaosylceramide) in the walls of blood vessels in all organs.

Clinical cases of low-symptomatic Fabry's disease in 2 girls (9 and 10 y.o.) with positive family history of the underlying disease are described.

Girl 9 y.o. is granddaughter and girl 10 y.o. is daughter of a male patient with Fabry's disease. Girls had complaints of reduced sweating and febrile acroparasthesia only.

Physical and sexual development according to age, the skin was clean.

Echocardiography, abdominal USE, pulmonary function test, cornea and fundus, ear, nose and throat examination were normal. Urine examination showed 1+ proteinuria both girls have. Glomerular filtration rates were normal. The lyso-Gb3 and galactosidase A levels analyzed in dried blood spots (DBS) by tandem mass spectrometry and molecular genetic analysis was performed. An increase lyso-Gb3 concentration and a change in the nucleotide sequence c.983G>C, leading to the replacement of p.G328A, described in the international database on HGMD mutations (CM 930337) in the hemizygous state has been identified.

Conclusion These cases confirm that it is very important that the pediatricians become aware of the importance of genealogical anamnesis findings and Clinical features of Fabry's disease, so they can participate in the identification of unrecognized patients.

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TYPICAL PROBLEMS OF PARENTS OF CHILDREN WITH CLEFT LIP AND PALATE

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