NS associated with c.153delT are particularly severe as demonstrated in our patient.

Testing of both parents was recommended as each parent of a child with autosomal recessive disease is generally considered to carry at least one copy of an aberrant allele. We found that her father has a pathogenic variant 153delT, p.(Gln52LysfsTer6), while her mother has no pathogenic variants. Genetic counseling emphasized that the genetic finding in our patient was not due to uniparental disomy and that the likely cause is de novo mutation in the maternal germinal cell. Although no specific treatment is available for Netherton syndrome, the first year of life has to be carefully managed, with rigorous care procedures that include multidisciplinary care. This approach should include careful monitoring and treatment of infections and hypernatremia, individual recommendations for calorie and protein needs for adequate growth, careful monitoring of allergic sensitization and dietary advice to optimise their nutritional intake, local skin treatment and supportive immunoglobulin therapy.

Following introduction of immunoglobulins, the patient had no infections during 2-years follow-up. However, despite intensive and individualized dietetic approach and nutritional treatment, our patient is still severely malnourished (WHO z-score: weight-for-age -2.49; length-for-age: -1.29; weight-for-length: -2.5).

160

FOLLOW-UP OF TWO CHILDREN WITH VERY LOW IGE FROM BIRTH

Helena Tesari Crnković*, Andrea Šimić Klarić, Marijana Tomić Rajić, Vlado Drkulec, Romana Gjergja Juraški, Mirjana Turkalj. General County Hospital Požega, Faculty of Medicine, J.J. Strossmayer University of Osijek

10.1136/archdischild-2021-europaediatrics.160

Background Immunoglobulin E (IgE) is best known for its association with allergic diseases. Very low IgE may be a marker for other immunodeficiencies. Deficiency or low IgE is defined with IgE concentration lower than 2.5 IU/ml. A majority of studies suggest a prevalence of 1-2.6 %.

The clinical significance of an isolated low IgE („selective IgE deficiency”) is unknown and is not currently included in the consensus classification of Primary Immunodeficiency Diseases (PIDD). Low IgE is suggestive of a primary humoral immunodeficiency, like Common variable immunodeficiency (CVID). In recent studies a connection between low serum IgE and low levels of one or more classes of immunoglobulins are found. The children's group was characterized with a higher prevalence of asthma.

Both groups (children and adults) had significantly higher prevalence of chronic sinusitis, otitis media, autoimmune, and oncological diseases than controls. In our case report two cases of very low IgE are presented. Both of them have been followed up from birth.

Case presentation: a cohort of 72 children has been followed up from birth up to 10 years of age. In two of them a very low serum IgE was found. Serum IgE was assessed at birth from cord blood, and reassessed at 1, 2 and 10 years of age with specific IgE (sIgE) for standard palette of food and inhalant allergens. At 10 years of age skin prick test, measurement of fractional exhaled nitric oxide (FeNO) and spirometry have been conducted.

The girl had an unmeasurable level of IgE in cord blood, and also at 1 and 2 years, and her IgE at 10 years was 2.0

IU/ml. The boy's IgE in cord blood, and at 1, 2 and 10 years was 0.11, 1.0, 8.2 and 0.1 IU/ml, respectively.

They both had symptoms characteristic of asthma and allergic rhinoconjunctivitis throughout their childhood, with negative sIgE at 1, 2 and 10 years of age and no evidence of type I hypersensitivity. Their other immunoglobulin (IgG, IgM, and IgA) levels at 10 years was within reference range for age, and they showed no symptoms suggestive of immunodeficiency or autoimmunity.

Conclusion here we report of two cases with selective IgE deficiency in children who had been followed up from birth. Very low serum IgE may serve as a marker of immune dysregulation and autoimmunity, and should trigger appropriate investigation (immunoglobulin quantification). The children will be closely monitored.

161

52-WEEK LABORATORY SAFETY FINDINGS FROM AN OPEN-LABEL EXTENSION STUDY OF DUPILUMAB IN ADOLESCENT PATIENTS WITH ATOPIC DERMATITIS (LIBERTY AD PED-OLE)

Michael J Cork*, Benjamin Lockshin, Andrew Blauvelt, Zhen Chen, Faisal A Khokhar, Ashish Bansal, Randy Prescilla. University of Sheffield, Sheffield, UK

10.1136/archdischild-2021-europaediatrics.161

Objective No clinically significant changes in hematology/chemistry parameters were reported after clinical trials of dupilumab in adults and adolescents with moderate to severe atopic dermatitis (AD). Here, we report long-term laboratory outcomes in adolescent patients (aged ≥ 12 to < 18 years) from an ongoing long-term open-label extension (OLE) trial (LIBERTY AD PED-OLE; NCT02612454).

Methods Patients with moderate-to-severe AD who had previously participated in 1 of 3 prior dupilumab studies were enrolled in this OLE study. In March 2017, weight-based weekly dosing (2 or 4 mg/kg) was amended to a fixed-dose regimen (300 mg every 4 weeks), which could be up-titrated in case of inadequate clinical response at Week 16 as: patients < 60 kg, 200 mg every 2 weeks (q2w); patients ≥ 60 kg, 300 mg q2w. Concomitant topical corticosteroids and topical calcineurin inhibitors were allowed. Laboratory safety data for the adolescent cohort were evaluated (data cutoff: December 15, 2018) from OLE baseline (n = 299) to Week 52 (n = 105).

Results Hematology assessments at Week 52 demonstrated that mean (standard deviation [SD]) counts of eosinophils ($0.58 \times 10^9/L$ [0.52]), basophils ($0.01 \times 10^9/L$ [0.03]), monocytes ($0.46 \times 10^9/L$ [0.17]), lymphocytes ($2.17 \times 10^9/L$ [0.65]), neutrophils ($4.11 \times 10^9/L$ [1.58]), leukocytes ($7.35 \times 10^9/L$ [2.03]), hemoglobin (139.7 g/L [13.80]), and platelets ($283.5 \times 10^9/L$ [65.28]) remained within the normal reference range for this adolescent population. Similarly, chemistry parameters at Week 52 showed no clinically relevant changes from baseline as seen from mean [SD] levels of creatinine kinase (141.6 U/L [117.3]), albumin (45.9 g/L [3.0]), protein (72.5 g/L [4.7]), bilirubin (7.0 $\mu\text{mol/L}$ [5.5]), potassium (4.3 mmol/L [0.3]), alkaline phosphatase (160.1 U/L [92.0]), creatinine (57.2 $\mu\text{mol/L}$ [13.4]), blood urea nitrogen (4.2 mmol/L [1.2]), lactate dehydrogenase (186.1 U/L [39.7]), and glucose (4.9 mmol/L [0.7]).

Conclusions No clinically meaningful changes in hematology/chemistry parameters were seen in adolescents with 52 weeks