Methods The Illumina Trustight One Sequencing Panel was used for sequencing over 4800 genes known to be associated with a clinical phenotype and spanning 12 Mb of genomic content. The panel studied contains 125,000 probes based on the NCBI37/hg19 human reference genome. X-ray and MRI imaging were performed for fibular hemimelia.

Results The patient was born by elective cesarean section at 38 weeks with 2805 grams. He was the fourteenth pregnancy and ninth living baby of the 32-year-old mother. The infant’s birth length was 45 cm, and the head circumference at birth was 35 cm. Physical examination of the patient revealed medial angulation in the right lower extremity from the knee, clubfoot deformity in the foot, and polydactyly in the left foot. There was no associated facial dysmorphism nor other associated anomalies apart from polydactyly, Abdominal, hip, and transfontanel USG and echocardiography were normal. Fibular hemimelia was found in the patient on x-ray and MR imaging. PMM2 and MEFV gene mutations were found in the gene analysis.

The patient was consulted to the orthopedic unit. Although limb amputation was recommended by surgeons, we investigated possible alternatives. As a result, the patient was referred to an external center for a tibial lengthening procedure.

Conclusions Congenital disorders of glycosylation are a group of hereditary diseases and they may present with different extremity anomalies.

British Society for Rheumatology

Background Juvenile-onset systemic lupus erythematosus (JSLE) is a rare, multi-system autoimmune disease. SLE most frequently affects adults but onset is in childhood in up to 20% of cases. Where onset is in childhood, disease can be more severe, leading to significant associated morbidity and damage.

Objectives This British Paediatric Surveillance Unit (BPSU) study of JSLE aims to describe presenting features, classification criteria, initial management and disease damage in the UK and Ireland population.

Methods BPSU methodology was used to identify new diagnoses of JSLE in children <18 years of age between September 2017 and September 2019. Relevant adult clinicians (including adolescent and adult rheumatologists, dermatologists and nephrologists) were also asked to report cases using parallel methodology. Data was collected at diagnosis and at one year follow-up. Data was analysed descriptively. McNemar’s test was performed to compare sensitivities of different classification criteria.

Results 253 cases were reported with 131 incident cases included to date (122 exclusions: 60 duplicates; 26 diagnosed outside study period; 15 case definition not met; 19 no clinical information; 2 diagnosis removed at one year). Systemic Lupus International Collaborating Clinics (SLICC-2012) classification criteria were more sensitive for classifying patients with JSLE than American College of Rheumatology (ACR-1997) classification criteria (86.3% vs 75.6%, p<0.05). European League Against Rheumatism/ACR (EULAR/ACR)-2019