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.

Paediatricians with Expertise in Cardiology Special Interest Group

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EFFECT OF SERUM CHLORIDE AND SERUM SODIUM DERANGEMENTS ON DIURETIC RESPONSE IN CHILDREN ON CHRONIC FUROSEMIDE THERAPY

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Background Electrolyte disturbances are not uncommon in patients on chronic Furosemide therapy. The individual effect of both hyponatremia and hypochloremia has been studied in adults but not in the children with congenital heart disease and chronic congestive state.

Objectives We aimed at studying the occurrence of serum Chloride (Cl) and Serum Sodium (Na) abnormalities in children on prolonged Furosemide therapy and the possible relation of these abnormalities to the diuretic response in this population.

Methods The study included forty-five children, with congenital left to right shunts causing chronic congestive state which necessitated chronic Furosemide therapy. Patients in need to an increase in their Furosemide dose were recruited in the study. We assessed serum Cl and serum Na as well as parameters of diuretic responsiveness; net fluid output and change in body weight/40 mg Furosemide, and change in urinary Na/K ratio. These parameters were assessed initially and at day 3 after increasing Furosemide dose.

Results Median age of patients was 0.75 years, with 27 (60%) male patients and 8 (40%) female patients. According to serum levels of Cl and Na, patients were divided into four groups; isolated hyponatremia (15 patients, 33.3%), isolated

hypochloremia (9 patients, 20%), combined hypochloremia and hyponatremia (12 patients, 26.7%), and normal serum electrolytes (9 patients, 20%). Patients with combined hyponatremia and hypochloremia and those with isolated hypochloremia showed minimal clinical and radiological signs of decongestion as well as lowest changes in urinary Na/K ratio, fluid output and weight change/40 mg Furosemide on augmenting the diuretic dose, unlike the hyponatremic patients who had near normal parameters with no evidence of diuretic resistance.

Conclusions Both hypochloremia and hyponatremia are common in patients on prolonged Furosemide therapy. Hypochloremia is associated with a poor diuretic response, unlike isolated hyponatremia which does not seem to affect the diuretic response. Concomitant occurrence of hyponatremia and hypochloremia is associated with poor diuretic response as well which can be worse than that seen in isolated hypochloremia.

British Society for Rheumatology

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PROSPECTIVE UK AND IRELAND POPULATION-BASED STUDY OF JUVENILE-ONSET SYSTEMIC LUPUS ERYTHEMATOSUS

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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