were detected in bone marrow aspiration material. Liver fibrosis was detected on workup. The diagnosis was established as GD, using enzyme studies. Molecular investigation of the patient showed homozygosity for a double mutation [H255Q; D409H]. Enzyme replacement therapy was initiated and was given twice in total. The patient died as a result of portal hypertension and gastrointestinal bleeding at age 8 months. We report the first Turkish patient to have a homozygous double mutation [H255Q; D409H]. Apart from other cases with the same mutation, our patient's neurological involvement was minimal to none, while gastrointestinal pathology (liver fibrosis) was the main factor for deterioration and death.

24. Lysosomal disorders: others

A-024

A novel mutation described in a turkish patient with infantile neuronal ceroid lipofuscinoses

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Neuronal ceroid lipofuscinoses are a heterogenous group of neurodegenerative disorders which are autosomal recessively inherited and characterized by the intracellular accumulation of autofluorescent lipopigments in neurons and other tissues such as retina of the eye. Neuronal ceroid lipofuscinoses are characterized by progressive loss of acquired functions, intellectual disability, myoclonic epilepsy and decline in motor skills. A two year old girl was the first living child of consanguineous parents presented to our clinic with restlessness. Her development had been appropriate until the end of the first year of her life. At first she had an ataxic gait and then she completely lost her ability to walk at the age of 18 months. On physical examination she was conscious; she had significant truncal hypotonia and severe hypertonia in her extremities. Deep tendon reflexes were increased. She did not have a social smile or follow with her eyes. There was no organomegaly. Magnetic resonance imaging of the brain revealed diffuse cerebral atrophy and corpus callosum agenesis. Neuronal ceroid lipofuscinoses was considered as diagnosis, and mutation analysis revealed a novel mutation in the PPT1 gene (p.P238Cfs*56, c.712_713delCC).

29. Miscellaneous

A-025

Functional independence of Taiwanese children with Down syndrome

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Background: Information regarding the functional strengths and weaknesses of children with Down syndrome is important for early intervention programs, agencies providing family support, and educational services.
Methods: We used the Functional Independence Measure for Children (WeeFIM) questionnaire on parents or caregivers of 166 Taiwanese children with Down syndrome to assess their functional skills (101 males and 65 females; median age, 12.7 years; age range, 3.2 to 19.1 years).
Results: The mean total WeeFIM score was 101.2 out of a potential score of 126. One hundred and fifty-four children (93 %) were identified with full trisomy 21, 7 with mosaicism (4 %), and 5 with the translocation type (3 %). The mean total WeeFIM score of each type was 100.6, 111.9, and 102.4, respectively ($p>0.05$). The mean scores for three domains (self-care, mobility, and cognition) were 45 (maximum 56), 33 (maximum 35), and 23 (maximum 35), respectively. Performance was strongest in the mobility domain, but weakest in the cognition domain. The total WeeFIM scores and 18 sub-scores for these three domains all positively correlated with age ($p<0.05$).
Conclusions: The WeeFIM questionnaire may be useful for the monitoring of long-term response to interventions in these children, as well as in subjects with developmental disabilities.

A-026

A child with combined chromosomal abnormality, mitochondrial dysfunction and disordered cobalamin metabolism

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Background: Mitochondrial dysfunction manifests with multiorganic disorders mainly affecting CNS, heart, liver, and muscles.
Case report: 4 month-old child. Development delay: weakly holds her head, lethargy, mild paratrofia. History: first days of life - lethargy, dyspnea, diffuse hypotonia and hyporeflexia. No sucking reflexes. Ultrasound: increased echodensity or the brain, subependymal cyst on the left side 2 mm; operating oval window, open ductus arteriosus. Elevated echodensity of the liver, metabolic nephropathy. Intensive therapy was provided. Hypotension, developmental delay; hydrocephalic syndrome. Tendon reflexes were average on hands, on knees were torpid. Electromyography at ag 3 months: upper limb muscle function reduced due to nerve damage along trunks of the brachial plexus, decreased muscle contraction of the hip; central type hypertonicity. Neurosonography - expanded external cerebrospinal fluid spaces. Karyotype: 47,XX, +mar. In blood: lactate ↑2.69 mmol\l, ↑LDH 545.68U/L, ↓creatinine 22.14mkm/L. Gas chromatography of urine: modified Krebs cycle metabolites; ketosis, ↓ Vit B2, B5, B12. Metabolic therapy - ubiquinone, carnitine, vitamins. General condition improved.
Diagnosis: chromosomal abnormality (marker chromosome), mitochondrial dysfunction, metabolic cobalamin disorder.
Conclusion: combination of chromosome pathology with metabolic disorders. We need to investigate chromosomal aberrations of metabolic status to choose adequate therapy.

A-027

Diagnosis of familial mediterranean fever masked by symptoms of chronic pancreatitis

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Familial mediterranean fever (FMF) is the most common hereditary periodic fever syndrome in populations of Mediterranean origin (Arabs, Turks, Jews, Lebanese, Greeks, etc.). The frequency of heterozygous carriers of MEFV, responsible for the development of FMF, is more than 1/5 of the total population. Case report: 22-year-old Lebanese student of Kharkiv National Medical University complained of acute pain in mesogastrium, nausea and fatigue. Similar episodes had occurred two years earlier, and the diagnosis of chronic pancreatitis was made. Given the cyclical nature of pain and the Mediterranean origin of the patient, molecular genetic testing was performed. by a combination of restriction fragment length analysis and allele-specific hybridization methods (Tibnin Governmental Hospital). The patient is compound heterozygous

for the mutations M694I and M694V in MEFV exon 10. The mutations V726A, E1480Q, M680I, P369S, R408Q, A744S, M680Ib, R761H, R65, F479L, E167D, K695R, and I692del were excluded. FMF was diagnosed in this patient with symptoms of chronic pancreatitis. It is known that the severity of clinical signs may be decreased in heterozygous carriers of mutant alleles. However, given the risk of FMF-associated amyloidosis, rheumatologist clinical supervision and control of serum amyloid were recommended.

A-028

A case of a possible ciliopathy

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Introduction: Ciliopathies comprise a group of disorders associated with genetic mutations encoding defective proteins, which result in either abnormal formation or function of cilia. They can manifest as a constellation of features that include characteristically, retinal degeneration, renal disease and cerebral anomalies. Nephronophthisis the most frequent genctic cause of ESRD in the first three decades of life results from mutations in different recessive genes (NPHP1 to *NPHP11*).
Case Report: 13-year-old girl with a previous history of unclear endocrinopathy admitted for renal failure. She has mild psychomotor development delay, is small for age and later on has mild ataxia. Investigations showed growth hormone deficiency, mildly high lactate, severe pigmentary retinitis, muscle biopsy mild atrophy of type II fibres, slight predominance of type I fibres and very mild respiratory chain complex I deficiency (36.1 %). A renal biopsy revealed glomerular sclerosis and cystic degeneration of tubules suggesting a possible nephronopthisis.
Comment systic kidney diseases get increasingly complex and there is evidence for a genetic and proteomic network with mutations in multiple cilia-related genes The association of the ocular changes, endocrinopathy, a hypothetical nephronopthisis and ataxia led us to think that the primary disorder may be an unknown syndrome or a possible ciliopathy.