She is the first born of a 51-year-old father and 40-year-old mother, non-consanguineous parents, at full term, and has a history of early hypotonia, psychomotor developmental delay and failure to thrive.
Physical exam revealed arthritis of both ankles, the right knee and the MCP joint of the left thumb. We noted loose joints, high-arched palate and facial dysmorphism (hypertelorism, large protruding ears, flat nose and short philtrum). Body weight and height were at -3SD. Intellectual disability was mild.
Laboratory tests showed slightly increased ESR (35mm at first hour) with positive ANA (>1/1000) and negative RF and anti-CCP. Karyotype is 46XX with deletion of the short arm of chromosome 18. Both parents had normal Karyotypes. MRI of the brain showed cortical atrophy and periventricular leucomalacia.
Ophthalmic screening detected bilateral anterior uveitis.
The patient was treated by AINS and methotrexate along with topical ophthalmic steroids. 6 months after treatment, the child had complete regression of uveitis and arthritis.
Results: This case of de-novo 18p deletion is associated to oligoarticular JIA with uveitis. According to our knowledge, there have only been 2 published cases of juvenile inflammatory arthritis with 18p deletion although 5 other cases were reported in association to other chromosome 18 abnormalities (deletion of long arm and ring). Few reports of adult-onset arthritis with monosomy 18p are available.
The exact explanation of this association is still unclear. As gene loss is a major factor in the pathogenesis of inflammation, this presentation might be related to deletion of PTPN2, a gene that has been associated to rheumatoid arthritis. PTPN2 encodes Protein Tyrosine Phosphatase Non-receptor type 2 and is located in the short arm of chromosome 18. PTPN2-deficiency stimulates T follicular helper cell and B cell responses thus promoting autoimmune reaction. Of the 67 genes in 18p, only around 12 (of which PTPN-2) are responsible of a phenotype when hemizygous. Further genetic studies are needed to better elucidate the role of deleted genes in the pathogenesis of inflammation in monosomy 18p.
Conclusion: In addition to skeletal deformities such as kyphosis and feet deformities, children with 18p deletion should be screened for inflammatory arthritis.

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AB0957 ATYPICAL PRESENTATION AND EARLY RECURRENCE OF KAWASAKI DISEASE IN A FEMALE INFANT: CASE REPORT

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Background: Kawasaki Disease (KD) is the second most common vasculitis in the pediatric population and the leading cause for pediatric acquired heart disease in developed countries. It is commonly diagnosed in the Mexican pediatric population, epidemiology in this country is not established, since cases are not usually reported to the healthcare system. The clinical features are quite variable, but diagnosis and prompt treatment will decrease morbidity and mortality. Coronary artery aneurysms are the most common complication, which represent the leading cause of acute coronary syndrome before 40 years of age. Recurrence of KD is estimated to be around 3% in Japanese patients and 1% in the United States, nevertheless, this data in Latin-American children is unknown. It usually affects patients before reaching 3 years old and within 2 years of the initial attack, presenting with an abrupt onset and higher complication rates, requiring aggressive workup, treatment and follow-up.
Objectives: To review an atypical presentation of KD and early recurrence in a 7-month-old female.
Methods: We present a 7-month-old female diagnosed with KD at 48 days old. She presented to the emergency unit with irritability, high and persistent fever and acholia. During workup, she was found with cholestasis and gallbladder hydrops. Negative CSF, blood and urine cultures were documented during hospitalization, and fulfilled the KD criteria. The cardiac ultrasound revealed coronary abnormalities: a RCA of 2.8mm (Z-Score +4.79), LCA of 3.3mm (Z-Score +5.69) and LAD of 2.4mm (Z-SCORE +4.25), which fit into classification 3 and 4, as small and medium aneurysms, according to AHA 2017. Immunoglobulin (2g/kg) and aspirin (80mg/kg) were administered and she was discharged 36 hours after the IVIg infusion, afebrile and with ambulatory follow-up, 30 days later the coronary abnormalities showed RCA of 2.0mm (Z-Score +2.3), LCA of 3.0mm (Z-Score +6), LAD 2.3mm (Z-Score +2.9). Six months later, she presented fever for 6 days, irritability and polymorphous rash. On physical exam BCGitis, non-suppurative conjunctivitis and pallor in hands and feet, elevated CRP and ESR, leukocytosis, thrombocytosis and sterile leukocyturia. Echocardiography reported RCA od 2.4mm (Z-Score +4.8), LCA of 3.1 (Z-Score +4.2) and LAD of 2.3mm (Z-Score +3.2), diagnosing KD recurrence, admitting the patient for IVIg and aspirin administration.
Results: The patient was treated with IVIg and aspirin. Follow-up by Cardiology determined improvement of Z-Scores. Recurrence occurred with worsening of the cardiac abnormalities. Cardiac prognosis is importantly affected due to the atypical age, vascular abnormalities and repeated vasculitic process. Rheumatologic consult should be considered since disease like Takayasu Arteritis, Polyarteritis Nosa and ADA2 deficiency need to be ruled out.
Conclusion: KD needs prompt diagnosis and treatment due to the potential consequences when delayed. Clinical suspicion is important due to the possible atypical presentation. As with this patient, age, gender and presentation are not exclusive. Despite adequate treatment, recurrence and worsening of the cardiac abnormalities occurred. Both KD events before 1 year old and with atypical presentations. Rheumatologic and cardiac follow-up need to be stringent through lifetime, to determine pharmacologic treatment, as ell as physical activity and reproductive counseling.

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AB0958 PEDIATRIC BEHCET'S DISEASE WITH SINUS VENOUS THROMBOSIS: THREE CENTER EXPERIENCE FROM TURKEY

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Background: Behçet's disease (BD) is an autoinflammatory disorder which may present with multi-systemic involvements including the vascular, cutaneous, articular, gastrointestinal, and/or central nervous systems (CNS). CNS involvement appears in two main groups: parenchymal or non-parenchymal. Neurological symptoms in children and adolescents can be confused with many other disorders and may be the initial symptom of BD.
Objectives: To report our experiences of the juvenile Behçet's Disease patients with cerebral venous sinus thrombosis (CVST) and to review previous studies reporting the clinical characteristics and outcomes of Juvenile Behçet's Disease with CVST.
Methods: The patients who met Pediatric Behçet's Disease (PEDBD) classification criteria for juvenile Behçet's Disease from 3 referral centers in Turkey were reviewed retrospectively. Disease activity was assessed by BD current activity form (BDCAF). A systematic review of literature of all published data was conducted.

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Results: The study group consisted of 12 juvenile BD patients with CVST. At the time of CVST diagnosis, the most common symptom was headache (%100), followed by vomiting (25%), blurred vision (16.6-7%), and disturbances in eye movements (16.7%). Six (50%) patients presented with sinus venous thrombosis as an initial symptom. Transverse sinus was the most frequently affected sinus (9/12, 75%) followed by superior sagittal sinus (8/12, 66.6%) and sigmoid sinus (1/12, 8.3%). The median (minimum-maximum) BDCAF was 6 (5-8). Four children (33.3%) had another venous thrombosis apart from CVST. All patients received pulse methylprednisolone for three consecutive days continued with oral prednisolone. Steroid treatment was tapered and discontinued minimum in six months. Eleven patients received azathioprine concomitant to steroid treatment at the time of CVST. All the patients received anticoagulant therapy concomitantly. Only one patient had relapse. Median (min-max) follow-up period was 4 years (1-10). In the literature review, we identified nine articles, describing 35 pediatric CVST patients associated with BD. Thirty patients achieved remission, while five patients had residual neurologic deficit.
Conclusion: Further multicenter studies with more patients and prospective follow-up may help us to understand the whole spectrum in these patients.

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AB0959 EXCELLENT RESPONSE TO TOCILIZUMAB IN PEDIATRIC ONSET REFRACTORY TAKAYASU ARTERITIS: CASE SERIES OF THREE PATIENT

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Background: Takayasu arteritis (TA) is a idiopathic and rarely seen chronic systemic vasculitis that affect the aorta and its major branches. Stenosis, occlusion and aneurysms may develop in large arteries in the setting of granulomatous panarteritis (1). The first purpose in patients with TA after diagnosis is to prevent the progression of vascular lesions with medical treatment. Used as a first choise in medical treatment, steroids and steroid-sparing immunosuppressive agents, may sometimes fail to prevent to progression of the disease. Interleukin (IL)-6, which synthesize from activated dendritic cells, is one of the main cytokine in the development of panarteritis in TA. Tocilizumab (TCZ), an anti-IL-6 receptor antibody, has been shown to be used as an effective treatment in many refractory adult TA patients (2).
Objectives: We have presented our experience with TCZ treatment in three children with refractory TA.
Methods: We reviewed three cases of childhood TA diagnosed between 2016-2018. These patients were successfully treated with TCZ that started due to the refractory disease.
Results: The first patient, 15 year-old girl, presented with fever, headache and abdominal pain. Physical examination showed systolic murmur in interscapular area and abdominal tenderness. The C-reactive protein (CRP) level was 4.7 mg/dL (n: <0.5 mg/dL) and the erythrocyte sedimentation rate (ESR) was 78 mm/h (n: <20 mm/h). Doppler ultrasound imaging showed wall thickening in bilateral carotid arteries. The angio-CT imaging revealed that 70% stenosis in the bilaterally renal arteries and partial stenosis in the mesenteric artery. Methylprednisolone treatment was started (30 mg/kg/d x 3 days). Maintenance treatment consisted of prednisone (1 mg/kg/d), methotrexate and azathioprine. Her complaints improved rapidly and the acute phase reactants decreased. However, the patient relapsed while tapering the corticosteroid.
The second patient was diagnosed with TA in 2012 after the complaint of chest pain and finding of hypertension. MRI angiography revealed wall irregularities and stenosis in the descending aorta and dilatation in the ascending aorta. CRP and ESR levels found high. The patient was followed up with glucocorticoid and azathioprine treatment. After four years of follow-up, relapse of the disease was observed with symptoms of fever, chest and back pain.
The third patient presented with fever, abdominal pain, arthralgia in the left elbow and arthritis of the left knee, and was diagnosed with TA two years ago. His CRP and ESR levels were found elevated. The angio-CT imaging revealed that 70% stenosis in the 4 cm segment of the abdominal aorta, 70% stenosis in the right renal artery, 50% stenosis in the left renal artery and 90% stenosis in the origin of celiac trunk and superior mesenteric artery. Despite the given bolus methylprednisolone and methotrexate therapy, there had not seen any improvement in disease activity.
Tocilizumab was started at 8 mg/kg monthly in all three patient and they achieved complete clinical and laboratory remission.
Conclusion: These cases demonstrate that the TCZ treatment has been shown to be a successful option in TA patients resistant to conventional immunosuppressive therapies.
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Informed consent form was obtained from patients and their legal guardians

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AB0960 THE HELIOS (HACETTEPE UNIVERSITY ELECTRONIC RESEARCH FORMS) REGISTRY: USE OF BIOLOGIC DRUGS IN AUTOINFLAMMATORY DISEASES

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Background: Autoinflammatory diseases (AID) are characterized by a dysregulation of innate immunity leading to uncontrolled inflammation. The treatment in AID is critical to control the disease activity, to prevent complications, and to improve the health-related quality of life. Biologic drugs have revolutionized the treatment and outcomes in AID.
Objectives: Herein we aim to present the clinical characteristics of children to whom biologic drug therapy was initiated for the management of AID.
Methods: A web-based registry called the Helios Registry (**Hacettepe univErsity eLectronIc research fOrmS) has been formed to evaluate the data of all children on biologic treatment. We have been enrolling patients since August 2018 retrospectively and prospectively. We have analyzed the data about the general characteristics of the patients, treatment, the biologic drug used, and adverse effects. Only the patients with the following diagnoses were included: systemic juvenile idiopathic arthritis (SJIA), familial Mediterranean fever (FMF),** cryopyrin associated periodic syndrome (CAPS), and chronic recurrent multifocal osteomyelitis (CRMO).
Results: Of 60 patients included, 19 had FMF (31.7%), 24 had sJIA (40%), 10 had CAPS, (16.7%), and 7 had CRMO (11.7%). Their median age was 10.7 (2-20) years old and disease duration was 2.8 (0-6) years, at the time of biologic drug initiation. 58.3% were currently on canakinumab, 20% anakinra, 10% tocilizumab, 10% etanercept, and 1.7% adalimumab. 63.3% of our patients had previously used at least one other biologic drug. The rate of glucocorticoid use before biologic treatment was 56.6%. The median duration of glucocorticoid treatment after initiating biologic drugs was 7.4 months. 56 (93%) patients achieved remission on biologic therapy. There were 15 patients (25%) who received tuberculosis prophylaxis due to positive tuberculin skin test (diameter$\geq$10 mm) and there was no Quantiferon test positivity. Thirteen adverse events (AE) had been noted. 2 of them were serious events as anaphylaxis due to tocilizumab infusions. The rest of the adverse events were mild thrombocytopenia (n=2), varicella infection (n=1), and local side effects (n=8). The median number of the infections per year was one and there were no death or malignancy.

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