

Rheumatology

Oral Presentation

Familial Mediterranean Fever; a 10 Year Follow up in Iranian Patients

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Objective: Familial Mediterranean Fever (FMF) is the most frequent periodic fever that is characterized by recurrent fever, abdominal pain, joint pain, skin rash and polyserositis. This disorder is seen sporadic in Iranian family. In this study, we report 10 years follow up and outcome of the disease in 85 patients from Iran.

Methods and Subjects: In a retrospective study, all referred patients during the last 25 years with a diagnosis of Mediterranean fever Rheumatology were studied. Following data were registered for all patients: early symptoms, initial laboratory results, follow-up period, drug doses, complications of disease and treatment. Infection, malignancy and rheumatologic diseases were ruled out in all patients.

Findings: Out of 91 patients who referred to rheumatology clinic with FMF diagnosis, 6 patients were excluded (due to incomplete follow up). Totally, 85 patients enrolled in this study including: 43 males (50.6%) and 43 females (49.4%). The mean age of patients was 13.0 (\pm 4.9) and the range 2.7 to 24.5 years. The mean age of patients at the first presentation was 49.2 (\pm 39.8) months (2 months to 15 years). The periodic symptoms was every 4.1 (\pm 2.1) weeks (range 1 to 8 weeks) before starting the treatment. The mean time of follow up was 8.9 (\pm 4.7) years (1 to 17.9 years). Regular and periodic symptoms have been mentioned in 64 patients (75.3%), but in others there are no regular periodic symptoms. Positive family history was recorded in 10.6% of patients, 6% in brothers and sisters and remaining in uncle or grandmothers. Common symptoms of FMF were: fever (91.8%), abdominal pain (84.4%), and joint pain (42.4%). Less common symptoms including chest pain, skin rash, and bone pain were recorded in 20%, 17.6% and 11.8%, respectively. Rare symptoms were vomiting (9.4%), pelvic pain (3.5%), neck pain and tension each one in 2.4% patients, and diarrhea and headache each one in 1.2% of patients. In

laboratory investigations, the mean of hemoglobin, platelets and ESR were 11.5 (\pm 1.4), 304000 (\pm 102077), and 29.3 (\pm 24.4), respectively. CRP in 20 patients (23.5%) was positive between 1 to 4+. Genetic studies was conducted in 17 patients that was normal in 47.1% of the patients. Out of 9 patients with abnormal genetic test, 5 patients (55.5%) had a mutation in the M694V gene, 3 patients (33.3%) a mutation in the M726A and 3 patients (33.3%) a mutation in the M680I gene. Two patients (2 / 22%) had a mutation in both M726A and M680I genes. All patients responded to Colchicine very good and there was no resistance to Colchicine in this study. In this study, 2.1% patients were symptom free with 0.25 mg, 15.3% under 0.5 mg, 74.1% with 1mg, 3.5% and 4.7% with 1.5 and 2 mg daily Colchicine. No serious complication due to treatments, or disease was recorded during follow up period.

Conclusion: According to our study, all symptoms and complication of FMF are controlled with proper treatment. There is not any Colchicine resistance or serious complication in our study. The frequent mutation was M694V and M726A genes in our patients, although 47% of patients had no known mutation genes.

Key Words: Familial Mediterranean Fever; FMF; Periodic fever; Colchicine; Genetic study

Oral Presentation

Childhood Systemic Lupus Erythematosus: Analysis of Clinical and Immunological Findings in 74 Iranian Patients

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Objective: The aim of this report is to describe the first instance of the clinical features of childhood lupus erythematosus. To define the pattern of disease expression in patients with childhood onset systemic lupus erythematosus (SLE).

Methods and Subjects: Patient Selection- We studied prospectively 74 patients with childhood-SLE who were seen consecutively either as inpatients or outpatients between 2000 and 2008 and who were followed up by the pediatric rheumatologist. All were met the American College of Rheumatology (ACR) revised criteria for SLE. In 74 the onset of disease, defined as the initial manifestation clearly attributable

to SLE, occurred before the age of 16, and they represented the childhood onset group described in this report. Laboratory investigations included complete blood count (CBC), erythrocyte sedimentation rate (ESR), Coombs' test, prothrombin time/partial thromboplastin time (PT/PTT). (ANA) were positive if higher than 1:160. Anti-dsDNA antibodies were positive if higher than 10U/ml. [Antiphospholipid antibodies (aPL) of the IgG and IgM. and complements (CH50, C3 and C4), urinalysis, 24/h/urine protein, BUN, Creatinine, chest X-Ray, ECG, and Echo cardiography]?. This is the first report of childhood systemic lupus erythematosus from the Iranian children. Childhood onset patients more often received prednisolone, hydroxychloroquine, azathioprine, methylprednisolone and cyclophosphamide pulps.

Findings: A fifteen-year retrospective analysis of the clinical features and survival of 74 Iranian children with systemic lupus erythematosus (SLE) was made. All the patients fulfilled the 1982 (ACR) revised criteria for SLE and had the disease at or before the age of 16 years. Sixty five (88%) patients from the childhood onset group were females and nine males (12%) (ratio female/male, 7/1). Mean age of this group at disease onset was 10 years (range 3-16). During the evolution of the disease, the childhood onset patients had the mode of presentation as follows: 55 patients (74%) had skin involvement, 35 patients (77%) had musculoskeletal involvement, 32 patients (43%) had renal disease, 25 patients (33%) had hematological abnormalities, 18 patients (24%) had pulmonary involvement, 13 patients (17%) had central nervous system involvement, and 12 patients (16%) had cardiovascular disease. Anemia was in approximately 44/74 (59%) of patients. Autoimmune thrombocytopenia purpura in 34/74 cases (45%), Leukopenia with lymphopenia was the presenting feature in 12/74 cases (16%). ESR >85 in 58/74 (78%) cases, and positive CRP(C-reactive protein) in 44/74 (59%) patients. Hematuria was the most frequent finding in these patients 35/74 (47%). Proteinuria was the second finding in our patients 32/74 or (43%). Raised BUN and creatinine was seen in 16/74 or (21%). The Coombs' test was positive in 16/74 children (21%), false positive VDRL in 12/74 patients (16%) with childhood-SLE. ANA positivity was detected in 72/74 (97%) of cases at presentation; the mean titer was >1:160 in all patients except 2 cases. All 2 children who were ANA-negative had at least a malar rash, oral ulcer, and associated with several mild manifestation. Anti-d DNA was positive in 62/74 (83%) patients. Antiphospholipid antibody was seen in 10/74 (13%) patients. 10% of patients with SLE would (were)?? be anti-Sm positive, low C3 (85%), low C4 (41%), and low CH50 complement (85%).

Conclusions: Childhood-SLE is not a common illness in the pediatric population. Although Childhood-SLE has been reported in children in first 10 to 20 years of life; it is rare in children under 5 years of age.

Key Words: Systemic lupus erythematosus, F-ANA, Malar rash

Oral Presentation

Spondyloepiphyseal Dysplasia Tarda with Progressive Arthropathy Mimicking Juvenile Chronic Arthritis; Report of 8 Cases

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Background: SED is one of a group of skeletal dysplasia (dwarfing condition) caused by changes in type II collagen. Two major types of SED are recognized, SED congenita and SED tarda. SED tarda is milder and late in onset, and appearance may be normal at birth. They present with short stature and symmetric joint pain caused by epiphyseal changes. A progressive "pseudo rheumatoid" arthritis has been described in some children with SED tarda.

Cases Presentation: This is a case series study including 8 cases from four families (5 girls and 3 boys). They were 9-17 years old and onset of clinical manifestations were 2-6 years old. Parents were first-degree relatives in three families. All of the cases were normal at birth. The most common signs were swelling, stiffness, range of motion limitation movements and contracture joints particularly knees, elbows and, PIP and DIP of both hands and feet. All of patients were short stature. (<fifth percentile curve). Also, patients had disproportionate trunk shortening. There were waddling gate in all of cases, kyphoscoliosis in 5, congenital coxavara in 2, chest deformity in 2 and varus valgus in 3. Thoracolumbar and pelvic x-ray showed pelvic flattening, characteristic vertebral posterior hump and vertebral fusion in 5 cases. Eye examination showed myopia only in 1 case. Cardiac examination, acoustic and intellectual evaluation were normal. Moreover, laboratory findings were normal in all of patients

Conclusion: SED tarda is a x-linked recessive disease (milder form is AD). Characterized by abnormal gate, short stature, stiffness and contracture of joints. The clinical presentation mimic JCA and all of the patients primarily treated as JCA. It should be considered as differential diagnosis of familial disorders with contractures of stiff joints and familial disorders of short stature.

Key Words: Spondyloepiphyseal dysplasia tarda; Short stature; Pseudo rheumatoid arthritis

Oral Presentation

Generalized Muscle Weakness in a Two yr Old Infant: An Atypical Presentation of Kawasaki Disease**Mohammad Radvar, MD**

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Background: Kawasaki disease (KD) is a form of idiopathic systemic vasculitis. Diagnosis is based upon specific clinical parameters. Cardiac manifestations explain the mortality rate. They can be reduced by early treatment using IVIG. Atypical onset of KD is a frequent problem, leading to diagnostic mistake and also can delay correct diagnosis.

Case presentation: Patient was a two yr old male infant admitted with fever, irritability and acute onset of generalized muscle weakness, that started from previous day, so he could not sit or stand. In 4th day of admission severe nail peeling first in hands and then in toes appeared. IVIG and high dose ASA administered for patient. One day later patient's fever subsided and in 10th day of admission patient discharged while he could sit and stand, but had difficulty in running. In outpatient followup about 2 week later he could also run well.

Conclusion: Acute onset of generalized muscle weakness and inability to sitting and standing is an atypical presentation of KD in infants, and in any infant with this symptom and compatible laboratory data, we must consider KD as a likely diagnosis.

Key Words: Kawasaki disease; Atypical Kawasaki; Muscle weakness

Oral Presentation

Comparison of sTREM-1 in Serum and Synovial Fluid in Children with Arthritis in Mofid Children's Hospital**Reza Shiari¹, MD, Ph.D; Mahdieh Musavi², MD**

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Objectives: sTREM-1 is a member of Ig super family that produced by monocytes. The aim of this study was to determine the level of sTREM-1 in sera and synovial fluids of children with inflammatory arthritis.

Material and Subjects: This is a case-control study. In case group serum and synovial fluid

samples were analyzed for level of sTREM-1 (n=27). Children with arthritis were divided in 3 groups: JRA(n=18), and other inflammatory arthritis e.g. AS, HSP, reactive (n=9). In control group serum samples were analyzed for sTREM-1 level (n=24 healthy controls).

Findings: Mean of serum level of sTREM-1 in case group was 124 pg/ml and 84.5 pg/ml in control group (p-value: 0.008). Mean of sTREM-1 level in synovial fluid was 160.5 pg/ml (increased). In this study, there wasn't any correlation between sTREM-1 level and other acute phase reactants (ESR, CRP, etc). In case group 56% had increased levels of sTREM-1 (>83 pg/ml). Children with recurrent arthritis had increased levels of sTREM-1.

Conclusion: Increased level of sTREM-1 in serum and synovial fluid is not limited to infections and also increased in inflammatory processes e.g. JRA, HSP, Increased level of sTREM-1 in serum was parallel to increased level of sTREM-1 in synovial fluid. Very high levels of sTREM-1 in children with recurrent arthritis suggests that sTREM-1 might be a prognostic factor in children with JRA.

Key Words: sTREM-1; Inflammatory arthritis; JRA

Oral Presentation

Seasonality of Kawasaki Syndrome and Coronary Artery Disease**Farah Sabouni^{1,2}, MD; Setareh Mamishi^{1,2}, MD; Farzad Ferdosian², MD**

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Kawasaki disease exhibits a distinct seasonality, and short-term changes in weather may affect its occurrence. Seasonality was bimodal with peaks in January and June/July and a nadir in October. This pattern was consistent throughout Japan and during the entire 14-year period. Some years produced very high or low numbers of cases, but the overall variability was consistent throughout the entire country. Admissions for Kawasaki disease in Chicago are seasonal. Clustering is seen during the winter months in India. In our studies, on seasonality of Kawasaki disease in Iran admissions for Kawasaki disease were seen during the spring and summer and we revealed most of our patients with coronary artery disease in the winter. Atypical changes in weather affect the occurrence of Kawasaki disease and are compatible with a link to an infectious trigger. Seasonality represents a rich area for future research.

Key Words: Kawasaki disease; Coronary artery aneurysm; Season

Oral Presentation

Unusual Presentation of Henoch– Schönlein Purpura in Iranian Patients; A Case Series

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Objective: Henoch–Schönlein Purpura (HSP) is the most common vasculitis in children. Etiology of the disease is unknown. It is classified in leukocytoclastic vasculitis and it is an IgA dependent. The common presentation in this disease is coetaneous involvement (palpable purpura), arthritis or arthralgia, gastrointestinal involvement (abdominal pain or occult blood in the stool) and renal involvement (microscopic Hematuria).

Methods and Subjects: In a prospective study during September 2008 to August 2009, all patients who had HSP diagnosis enrolled in this study. CBC, ESR, CRP, FANA, U/A, BUN, Cr and abdominal ultrasound and biopsy were done in all patients. Uncommon features of disease were selected and analyzed. Skin biopsy and other necessary diagnostic such as testicular Doppler ultrasound, brain CT scan were performed. Data was analyzed by SPSS software (version 16).

Findings: Out of 29 patients 37.9% was female and 62.1% were male. Mean age of patients was 6.5(± 3.9) years. Spring and winter were common seasons (42.3% and 30.8%, respectively). Clinical symptoms were skin involvement in all patients (100%), gastrointestinal involvement in 75.9% (100% abdominal pain, 22.7% positive occult blood, 9.1% massive and recurrent GI bleeding and 31.8% abnormal sonography), joint involvement in 69.0% of patients (65.0% arthralgia and others arthritis), testicular involvement in 50.0% of boys (66.7% swelling of testicles, 11.1% epididimo-orchitis, 11.1% penis inflammation, and 11.1% prepus swelling), central nervous system involvement in 13.8% (75% headache, and seizures and confusion each one 25%), and renal involvement in 10.3% (microscopic hematuria). Hypertension was not detected in our patients in acute phase of HSP. Average days of hospitalization was 8.0 (± 5.2) days. In 10 patients (38.5%) disease reactivation were recorded and 4 patients needed to readmission (2 patients in the

first week after discharge, a patient 3 weeks after discharge and a patient 2 months after discharge). Uncommon features in our patients was massive lower gastrointestinal bleeding in three episodes in 2 patients, Aute Hemorrhagic Edema of Infancy in 2 patients (6.9%), penis inflammation, prepus swelling and epididimo-orchitis each one in one patient and urticaria, prolong fever and arthralgia in one patient. Also, 17.2% patients had auricular inflammation. Recent patient was followed up with HSP diagnosis and after 3 weeks he was readmitted with acute abdomen presentation and intussusception in ultrasound. Patients with massive or recurrent GI bleeding was controlled by IVIg, methylprednisolon pulse and cyclophosphamide pulse. Patient with urticaria, prolong fever and abdominal involvement was treated with oral prednisolon for two months. All patients were followed up between 4 to 12 months and microscopic hematuria was seen after acute phase in one patient (3.8%).

Conclusion: Although HSP is usually a self limit disease in children but unusual and severe forms may be required to treatment. In these situations, other vasculitis should be considered as a differential diagnosis. Unusal and life threatening symptoms may be required to treatment with prednisolon and immunosuppressive drugs.

Key Words: Henoch–Schönlein Purpura; Abdominal pain; Testitis; leukocytoclastic Vasculitis; Vasculities

Poster Presentation

Investigation the Rate of the Frequency of Clinical, Laboratory Manifestations of Patients Affected by Kawasaki Disease and Hospitalized in Qom Children's Hospital

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Objective: Kawasaki disease (the main cause of acquired heart disease in children) is an acute febrile multisystem vasculities with neurological, pulmonary involvement, renal and heart complications. The cause of this disease yet is unknown. The main symptoms of the disease are fever, bilateral bulbar conjunctivitis, changes in the mucosa of the oropharynx, erythem or edema of the peripheral extremities, rash and lymphadenopathy. There is no special diagnostic lab test for kawasaki disease. If diagnostic and treatment performed timely, heart complication can be completely controlled. In this study, we investigated the rate of the frequency of clinical, laboratory manifestations of patients affected by Kawasaki disease and hospitalized in Qom Children's hospital.

Method and Subjects: A study of case series performed on the 33 children affected by Kawasaki

which were hospitalized in Qom Children's hospital, under treatment during years 2001-2006. The information analyzed by SPSS (11.5) software.

Findings: The sequences of important clinical and lab test manifestations of this disease are: Fever: 33 persons (100%), Conjunctivitis: 30 persons (90.9%), Erythema or edema of the peripheral extremities: 14 persons (42.4%), Rash: 26 persons (78.8%), Lymphadenopathy: 20 persons (60.6%), decrease of Hemoglobin: 18 persons (54.5%), thrombocytosis: 16 persons (48.50%), increase of ESR: 30 persons (90.9%).

Conclusion: According to the importance of the involvement of heart disease and the affected people which did not get good treatment, the necessity of careful training for general practitioners is needed; and more attention of the experts to manifest the Kawasaki disease especially atypical one is identified.

Key Words: Kawasaki; Vasculitis; Lymphadenopathy

Oral Presentation

Painful Hip in Childhood; A Clinical Overview and Imaging Approaches

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A painful hip is a common clinical symptom of numerous joint, bone and soft tissue diseases, and is a diagnostic challenge for the pediatrician and

consulting radiologist. From clinical point of view it is important to know whether the symptoms are acute with sudden onset or if the complaint is more chronic. Also other clinical and laboratory findings are needed to suspect an adequate clinical diagnosis. In accordance to the frequency and incidence the patient with a painful hip should be categorized in 3 age groups, namely 0-3, 3-10 and 10-15 years old. In the first years of life the most common finding is arthritis of the hip or trauma. In the second age group transient synovitis is the most common finding, followed by Perthes disease. Slipped capital femoral epiphysis (SCFE) and malignancies are most often observed in the third age group.

Several imaging modalities can be used to visualize the hip joint. Sonography is the modality of choice in screening patients with a painful hip, especially in acute and non-traumatic conditions. Joint effusion with or without synovial thickening is easily detected as well as the atrophy of the muscle. Traditional conventional radiography is used to image the pelvic bone including the hip joint. Other imaging techniques such as radionuclide studies, computer tomography (CT) and magnetic resonance imaging (MRI) have great impact for diagnosis, but they can not be used routinely for the screening because of inconvenience, costs and radiation dose. In a period of more than 25 years we have had the opportunity to observe a large number of children with different diseases which manifest clinically as a painful hip. The availability and experiences with all imaging modalities enabled us to develop a flow chart for adequate imaging of the painful hip, useful for clinical practice. Within scope of this presentation different clinical and imaging findings will be shown.

Key Words: Hip pain; Imaging; Limping; Sonography