THU0531 "BOTHERING TO LISTEN": ENABLING PATIENT AND FAMILY INVOLVEMENT IN CHILDHOOD UVEITIS HEALTHCARE

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Background: Childhood uveitis is a group of heterogenous, potentially blinding inflammatory disorders. Management is complex. There is growing recognition of the importance of actively involving affected children and their families in their own care. Co-designed interventions, developed through active involvement of staff and patients, can provide effective solutions to problems identified by those affected.
Objectives: To use findings from a patient and family discussion group to inform the co-designed development of health care processes and interventions.
Methods: Five children/young people with uveitis (age ranges 8 to 17), seven parent/carers of children with uveitis and four health care professionals attended a 90 minute discussion group. Main discussion topic was the identification of areas in need of interventions or support structures. Sub-topics were determined a priori using previous PPI and existent research (REFS). They comprised: Direct health care; Impact on families; School, education and peers. Responses were collated. Consent was taken for use of direct quotes from participants.
Results: We outline the areas identified by children and families:
Direct health care: Four interconnected areas were identified: (1) Transitioning to adult services, (2) peer support (health care services being a valuable site for identifying peers), (3) communication between care structures, and (4) education for families. With regards to family education, there was identification of the need for specific services or interventions around the communication of (4a) diagnosis, (4b) treatment, (4c) likely long term outcomes/prognosis, (4d) and the child's progress. There was also discussion on (4e) the formats used to communicate with families.
Impact on families: Participants discussed support around (1) family relationships, (2) the impact of systemic medication. They also discussed (3) a need for recognition of the changing nature of their lived experience as affected families over the disease course and the need for on-going psychologic support especially at presentation to help with acceptance.
School, education and peers
Participants discussed the need for support around: (1) the impact of treatment on school life, (2) communication with school professionals and peers, (3) impact of visual impairments, (4) managing the visibility/invisibility paradox (ie being made to feel different but also having a disorder which was not externally apparent), and (5) managing adolescence.
Conclusion: Through the above approach, we have identified a range of issues affecting our patients and their families. Our findings are similar to those of other groups (1, 2) These lived experiences will be used to inform the co-design of supportive services (patient leaflets, videos, website, psychology intervention) and research on the effectiveness of these interventions in improving the management of affected children and their families.

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Disclosure of Interests: Sandrine Compeyrot-Lacassagne Grant/research support from: Abbvie, Christine Twoney: None declared, Harry Petrushkin: None declared, Dhanes Thomas: None declared, Emily Robinson: None declared, Lucia Kossarova: None declared, Ameenat Lola Solebo: None declared
DOI: 10.1136/annrheumdis-2019-eular.7262

THU0532 RITUXIMAB FOR RAPIDLY PROGRESSIVE JUVENILE SYSTEMIC SCLEROSIS

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Background: Juvenile Systemic Sclerosis (JSSc) is a rare multi-systemic disease characterized by fibrous changes of the skin and internal organs [1]. Patients with "rapidly progressive SSc" usually present rapid development of skin induration and important organ damage [3], leading to poor prognosis [2, 3]. Recently Rituximab (RTX), a monoclonal antibody against the CD20 antigen on B cells, has demonstrated to be a promising therapy for adult patients with SSc [4, 5].
Objectives: We describe four pediatric patients with rapidly progressive JSSc treated with RTX.
Methods: Data on clinical, laboratory and instrumental parameters were collected from four patients with rapidly progressive JSSc treated with RTX for at least one year. Data were recorded at baseline and every 6 months after initiation of therapy. All patients underwent i.v. RTX therapy with four cycles (375 mg/m^2 every 2 weeks), at 3 months intervals. Oral prednisone (PDN, 0.5 mg/Kg/day) and oral mycophenolate mofetil (MMF, 500 mg/m^2/day) were administered between RTX pulses. Skin changes were assessed by MRSS, changes on muscles involvement by CMAS. Variations on BMI, pulmonary function tests (FVC, FEV1, DLCO) and cardiac involvement (LVEF, LVEDV, GLS) were expressed as% change from baseline. J4S was used to assess the overall disease severity [6].
Results: Four JSSc patients (3M, 1F), aged 8-17 years, entered the study. Three patients presented with prevalent cardiological involvement. One patient (Case 3) presented with severe pulmonary involvement. After one year RTX treatment, all patients showed significant decreased of number/duration of Raynaud Phenomenon attacks and 3 patients of cutaneous involvement. Two patients needed an implantable cardioverter defibrillator (ICD) because of episodes of severe ventricular tachycardia (VT). After 12 months of therapy one patient presented improvement LVEF (+19%) and J4S, the other showed a global cardiac improvement (LVEF +37%, LVEDV -18%) and J4S. Both underwent a second year-long treatment with RTX with no worsening of internal organs' involvement. Case 3 showed a significant improvement of the respiratory function (FVC +46%, FEV1 +33%, DLCO +30%) with decrease of J4S. Case 4 improved her arrhythmia and muscle strength (CMAS +17%). No major RTX-related side effects were reported.
Conclusion: Rapidly progressive JSSc still carries a high mortality rate. To the best of our knowledge this is the first case series of patients with JSSc successfully treated with RTX. Our experience, although in a small cohort, confirms the beneficial effect of this therapy on the life-threatening internal organ involvement, particularly on cardiac function.

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Disclosure of Interests: None declared
DOI: 10.1136/annrheumdis-2019-eular.7377

THU0533 IMPAIRED PLATELET FUNCTIONS IN PATIENTS TREATED WITH COLCHICINE

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Background: Colchicine has been used in the treatment of Familial Mediterranean Fever (FMF) since 1972. Apart from the inhibiting mitosis in all cells, colchicine has an anti-inflammatory effect by inhibiting activation and migration of neutrophils. Colchicine is a safe drug at recommended

doses, but it can cause rare side effects including hematological findings such as lymphopenia, thrombocytopenia and neutropenia.
Objectives: In this study we aimed to define the adverse effect of colchicine on platelet function and its clinical relevance.
Methods: A total of 220 FMF patients between June 2016-2017, followed at Hacettepe University Pediatric Rheumatology Department and were on colchicine treatment for at least one year, were included to the study.
Results: Among the selected 220 FMF patients, 100 of them (54% female) described hematological symptoms when questioned in detail. The mean age of these patients was 11.74 ± 4.86 years. The mean cumulative colchicine exposure was 5.7±3.8 years. The most common referral symptom was frequent epistaxis (79%) followed by easy bruising (69%), and menstrual disorder including prolonged or heavy menstrual bleeding (21.8% among female patients). Among these 100 patients, 36 of them had prolonged bleeding time and impaired platelet aggregation test. Patients who had abnormal platelet function tests (the group with abnormal bleeding time) were receiving higher colchicine doses (median 0.05 vs 0.03 mg/kg/day; p:0.001) compared to the patients who had normal platelet function tests (bleeding time normal group) However there were no significant difference in terms of cumulative colchicine exposure (median 6.5 vs 4.5 years; p:0.07) and total platelet counts (median 288500 vs 279000/mm3; p:0.61). Patients with abnormal platelet function tests also had more epistaxis (47% vs 7%; p<0.001) bruising (51% vs 3%; p<0.001) and dysmenorrhea (among female patients 100% vs 26%; p<0.001). Colchicine was not reduced in these patients and no life-threatening event was observed.
Conclusion: In our study, we have shown prolonged bleeding time for the first time in the literature. Colchicine may cause microtubule inhibition in platelets as well as in other cells and impair platelet function. Further prospective studies are needed to clarify the significance of this side effect.

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Disclosure of Interests: Özlem Şatırer: None declared, Selcan Demir: None declared, Erdal Sag: None declared, Armağan Keskin: None declared, Yelda Bilginer: None declared, Şule Ünal: None declared, Seza Özen Consultant for: Seza Ozen is receiving consultancy fees from Novartis, Speakers bureau: Roche
DOI: 10.1136/annrheumdis-2019-eular.7713

THU0534 NEW CLASSIFICATION CRITERIA FOR RECURRENT AUTOINFLAMMATORY DISEASES APPLIED TO AN INDEPENDENT COHORT: EXPERIENCE FROM THE JIR COHORT DATABASE

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Background: New classification criteria for the inherited periodic fever syndromes (TRAPS, FMF, MKD and CAPS) have recently been developed during a Consensus Conference held in Genoa in March 2017.
Objectives: The aim of our study was to compare these new classification criteria for monogenic recurrent fever syndromes with the diagnoses of clinicians. For this purpose we used the JIRcohort database, an international platform gathering data of patients with pediatric inflammatory disease.
Methods: The Genoa classification criteria were applied to all the patients, then compared to the clinical diagnosis of the treating physician. As patient diagnosis could be confirmed or suspected, the patients could have up to two diagnoses. Classification criteria relied on genetical HRF pathogenicity classification. Finally, criteria performance were assessed by firstly determining sensitivity and specificity and secondly analyzing true positive, false positive and false negative patients.
Results: 455 patients included to the JIRcohort database with a recurrent fever syndrome were enrolled to the study.
CAPS: The analysis of the performance of the CAPS criteria showed sensitivity of 60% and specificity of 98%. 14 patients fulfilled Genoa CAPS classification criteria, with 6 true positive and 8 false positive patients. Patients with confirmatory genotype always fulfilled classification criteria. 4 patients, who carried heterozygous mutations, were false negative.
TRAPS: The analysis of the performance of the TRAPS criteria showed sensitivity of 100% and specificity of 98%. 22 patients fulfilled Genoa TRAPS classification criteria, all true positive patients with confirmatory and non-confirmatory genotype. 5 were false negative with 4 patients with non-confirmatory genotype.
FMF: The analysis of the performance of the FMF criteria showed sensitivity of 96% and specificity of 89%. 118 patients were true positive while 35 were false positive patients. True positive patients were all patients with confirmatory genotype, patients with non-confirmatory genotype and no mutation. 37 were false positive patients. 5 patients were false negative with 3 patients with non-confirmatory genotype.
MKD: The analysis of the performance of the CAPS criteria showed sensitivity of 64% and specificity of 66%. 7 patients with confirmatory genotype were true positive patients. 148 were false positive patients with 44 patients diagnosed TRAPS, FMF, CAPS while the rest were SURF. 4 were false negative including 3 patients with non-confirmatory genotype.
Conclusion: This study is the first Genoa criteria evaluation among a cohort of patients seen with recurrent fever. This descriptive study shows tremendous performance Genoa criteria for patients with confirmatory genotype and help classifying patients with non-confirmatory genotype. On the other hand, those classification criteria were less performant when patients did not display at least one gene mutation. Therefore, Genoa classification criteria for TRAPS outperformed the others, because mandatory genetical screening. This study also highlights permissive criteria for clinical CAPS, FMF and MKD. The implementation of biological criteria in MKD would improve MKD criteria.
Disclosure of Interests: : Glory DINGULU: None declared, Sophie Georgin-Lavialle Consultant for: novartis, sobi, Speakers bureau: novartis, sobi, Isabelle Koné-Paut Grant/research support from: SOBI has supported drug product (anakinra) for the presented study, Consultant for: SOBI, Novartis, Pfizer, Abbvie, UCB, CHUGAI, ROCHE, Pascal Pillet: None declared, Anne Pagnier: None declared, Etienne Merlin: None declared, Daniela Kayser: None declared, alexandre belot: None declared, Michael Hofer Grant/research support from: Sobi, Consultant for: Novartis, Véronique Hentgen Consultant for: SOBI, Novartis, Abbvie, Speakers bureau: Novartis
DOI: 10.1136/annrheumdis-2019-eular.7473

THU0535 NATURE AND IMPACT OF THE FRENCH NETWORK RESRIP ON SCHOOLING, FOR CHILDREN WITH CHRONIC INFLAMMATORY RHEUMATISM

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Background: Pediatric chronic inflammatory rheumatism (CIR) has a significant impact on daily life, especially on schooling, that can lead to child's drop-out. In this context, a French paediatric health network, RESRIP (Réseau pour les Rhumatismes Inflammatoires Pédiatriques), has been created in 2014 for children with CIR and living in the Ile-de-France region. Patients are included in this network if they meet certain criteria such as the need to find trained health professionals close to their home or for support because of adverse social conditions or difficulties with their schooling. If the criteria are fulfilled, the patient and his family participate in an intake interview which allows RESRIP to understand the patient's need and to set up targeted actions.
Objectives: In our study, we aim to describe and evaluate RESRIP's role on improving school attendance and tackling absenteeism
Methods: A descriptive retrospective study was performed regarding the support provided by RESRIP with respect to patients' schooling and education professionals. Rates of non-attendance were collected at inclusion

and every 6 months from 2014 to 2017, through a standardized auto-questionnaire.
Results: 278 patients (M/F: 0.29) aged 12.7 years on average (± 4.9, range 2-21) were taken care of between 2014 and 2017 by RESRIP. Juvenile idiopathic arthritis (JIA) (n = 142), connective tissue disease (n = 49) and auto-inflammatory disease (AID) (n = 32) are the 3 main pathologies covered. Among the 278 patients, twenty-one percent of patients needed academic support when entering the network, including: 37% in the JIA group, 34% in the connective tissue group and 18% in the AID group. Educational assistance was set up for all patients with school difficulties at inclusion but also for all the patients during their follow-up.
Patient Support: 178 Individual Action Plan (IAP) were implemented by RESRIP. Twelve patients benefited from additional time for their school exams and 10 were allowed to pass the baccalaureat (French final college exam) over two years instead of a year. In thirty patients, sports education has been adapted. Forty-one MDPH files (Departmental Houses for the Disabled) were produced to enable the establishment of a school life assistant (AVS), teaching materials and/or technical aids. Finally, 8 patients received home schooling assistance through Home Learning Assistance Services (SAPAD).
Education professional support: RERSIP established partnerships with the French National Education (FNE) and the SAPAD (home tuition service for ill children). For the FNE, RESRIP provided school doctors or nurses with: 8 continuing medical training and 25 personal interviews to explain the pathologies. In addition, 5 multidisciplinary meetings, within the institutions were organized, for 5 patients with social integration difficulties and stigmatization. RESRIP actions, has allowed a significant decrease in school absenteeism between 2014 and 2017: 3.2 days per year on average at day 1 to 0.5 days per year on average ($p < 0.05$) at 42 months.
Conclusion: Over time, a better understanding of the impact of chronic illness over school and education professionals, has allowed RERSIP to improve its support. The result is a notable decline in school absenteeism and in unjustified physical education exemptions. Several projects are underway: The development of a standard IAP available for doctors, the set-up of a partnership with hospital school, willing to help for the implementation of home school support courses in addition to SAPAD.
Disclosure of Interests: : Perrine Dusser: None declared, Chrystelle Hascoet: None declared, Isabelle Koné-Paut Grant/research support from: SOBI has supported drug product (anakinra) for the presented study, Consultant for: SOBI, Novartis, Pfizer, Abbvie, UCB, CHUGAI, ROCHE, Linda Rossi-Semerano Grant/research support from: Roche
DOI: 10.1136/annrheumdis-2019-eular.7122

THU0536 LONG TERM IMMUNITY IN A PAEDIATRIC COHORT OF PATIENTS WITH RHEUMATIC DISEASES FROM A TERTIARY HOSPITAL IN SPAIN

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Background: Pediatric patients with rheumatic diseases (RD) are at increased risk of infections. Vaccines have been proved to be very effective to prevent them. Nevertheless the long-term immunity after vaccination remains quite unknown in this group.
Objectives: To comapre the long term seroprotecction in pediatric patients with rheumatic diseases who recieved measles, rubella, mumps, tetanus, diphteria, hepatitis B, hib and meningoccocus C vaccination acording to the routine immunization schedule in Spain, and healthy children.
Methods: We designed a cross-sectional study including consecutive pediatric patients with RD who attended the rheumatology clinic and healthy children older than 10 y.o. The administered vaccines, treatment and pathology of each patient will be recorded. Their antibodies titters against each antigen were quantified and compared to healthy children.
Results: 60 patients (median age 13 y.o IQR 10.2-12-7) and 15 healthy children (mean age 11.3 y.o. IQR 11.3-12.7) were included. In the patients group 62% were female, and 35% in healthy. Diagnosis: 85% Idiopathic juvenile arthritis, 18% Lupus or juvenile dermatomiositis. 46% had received biologic treatment sometime. Seroprotection rate was (patient vs control): measles 85%% vs 81%%, rubeola 78.8% vs 73.3%, parotiditis 84.7% vs 66.7%; VHB 27.% vs 10.5%, diphteria 89.5% vs 81%, tetanos 64.9% vs 61.1%. Hib 42% vs 53%, pneumococo 92.4% vs 100%, meningococcus C 12% vs 11%. $p>0.05$
Conclusion: No statistical differences were detected, althought the scarce number of control subjects might have infuenced this result. There is a tendence towards a lower antibody persistence anti-Hib in patients compared to healthy children. The response to life virus vaccine and tetanus seem to be as good as in healthy chidren.
Disclosure of Interests: : Laura Fernández Silveira: None declared, M.J Gimenez: None declared, E Andreu-Alapont: None declared, Patricia Falomir-Salcedo: None declared, E Serrano-Poveda: None declared, M.I. Gonzalez-Fernández: None declared, B Lopez-Montesinos: None declared, Inmaculada Calvo Grant/research support from: received research grants from Pfizer, Roche, Novartis, Clementia, Sanofi, MSD, BMS and GSK, Consultant for: Advisory boards: Novartis, AbbVie, Speakers bureau: AbbVie, Roche, Novartis, SOBI
DOI: 10.1136/annrheumdis-2019-eular.6378

THU0537 ANGIOMATOID FIBROUS HISTIOCYTOMA MIMICKING SYSTEMIC JIA VIA MUTATION-DRIVEN IL-6 PRODUCTION

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Background: Angiomatoid Fibrous Histiocytoma mimicking Systemic JIA via mutation-driven IL-6 production: Angiomatoid Fibrous Histiocytoma (AFH) is a rare tumour associated with mutation-driven production of Interleukin-6 (IL-6) which causes a systemic inflammatory picture similar to Systemic Juvenile Idiopathic Arthritis (sJIA).
Objectives: A previously well 6-year old girl was referred with 6 weeks of abdominal pain, nausea, weight loss, night sweats, & lethargy.
Examination was unremarkable apart from a 2cm lump in the right popliteal fossa. This had been reported on USS 2 months earlier, at an external hospital, to be a Sebaceous cyst. There was no rash, organomegaly, lymphadenopathy, or synovitis.
She had persistent recurrent fevers of >39C, but not in the classical quotidian pattern of sJIA.
Methods: Bloods showed persistent Hb <70, platelets >600, ESR >100, & CRP >200.
Autoantibody & full infection screens were negative.
Urine HMMA:creatinine ratio was minimally raised at 7.2 (normal 1.8 - 5)
Faecal calprotectin, upper & lower GI endoscopy were normal.
Bone-Marrow Aspiration was reported as being highly reactive but with no malignancy seen.
Whole-body STIR MRI was reported as normal, but repeat localised USS &MRI of the knee showed a 27x18x21mm well circumscribed, mixed cystic/solid lesion with marked vascularity. The lesion was then biopsied, and subsequently excised
Plasma IL-6 levels were significantly elevated at 46.7pg/ml (normal range: 0-2), but normalised after excision. TNF and IL1b levels were normal
Results: Initial biopsy and FISH analysis confirmed the diagnosis of AFH with an abnormal EWSR1 signal pattern and classical EWSR1-CREB1 fusion. She went on to have a full excision, and staging investigations were negative.
AFH is rare (<0.3% of all soft-tissue tumours) and arises in the deep dermis/subcutis of the extremities of children & young adults. It has been regularly described in the axilla & popliteal fossa. It has a high (15%) local recurrence rate but rarely metastasises.
In >90% of cases AFH is associated with a characteristic translocation: t (2:22) (q33:q12). This forms the fusion gene EWSR1-CREB1 which in turn leads to continuous activation of CREB1. The promoter region of IL-6 has a CREB1 binding site thus causing IL-6 over-production and leading to the paraneoplastic syndrome.
Conclusion: Paraneoplastic immune dysregulation is a rare, but important cause of symptoms often otherwise associated with rheumatic diseases. Careful physical examination, and liaison with oncology teams, is needed, particularly before treatment with steroids or IL-6 inhibitors such as Tocilizumab are considered
Disclosure of Interests: : Mark Friswell: None declared, Stephen Owens Consultant for: paid a consultancy fee by Chimerix to act as an advisor on a study of adenovirus in transplant patients, Gail Halliday: None declared
DOI: 10.1136/annrheumdis-2019-eular.309