Figure 1. Relapse free survival of the four different IgG4-RD phenotypes.

Conclusion: Clinical phenotypes of IgG4-RD reflect differences in epidemiological features and prognostic outcomes.

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SAT0529

CLINICAL CHARACTERISTICS AND TREATMENT PATTERNS IN A PATIENT GROUP WITH INTERSTITIAL LUNG DISEASE.

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Background: Diffuse interstitial lung disease (ILD) is frequently associated with connective tissue diseases (CTD) and is one of the main causes of morbidity and mortality in these patients. Recently, the concept of Interstitial Pneumonia with Autoimmune Features (IPAF) has been defined to characterize ILD associated with systemic manifestations limited to subtle serological and clinical autoimmune abnormalities and not fulfilling the international criteria for the diagnosis of a given CTD.

Objectives: The objective of this study is to describe the clinical, serological and radiological characteristics, as well as the treatment patterns of patients with ILD referred to a Rheumatology Service for suspected CTD

Methods: Observational, cross-sectional study of 43 patients with ILD referred for evaluation to the medical consultation of CTD of the Rheumatology service at the Reina Sofia Hospital. Patients were classified as patients with defined CTD, patient with IPAF and patients with other types of pneumopathy. We conducted a descriptive study of all patients and compared the clinical-analytical-radiological characteristics and treatment patterns of the first two groups.

Results: Of the 43 patients, 67.40% were women with a mean age at diagnosis of 65.65 (10.42) years and 53.50% of smoking patients. Of the total of patients, 16 (37.2%) were included in the CTD group, 17 (39.5%) met criteria for IPAF and 10 (23.3%) had another type of pneumopathy. In the CTD group scleroderma was the most frequent disease (6/16), followed by inflammatory myopathy (4/16), Sjögren's syndrome (3/16), rheumatoid arthritis (2/16) and polymyalgia rheumatic (1/16). In this group of patients, the most common symptom was Raynaud's phenomenon (RP) (7/16), followed by arthritis (7/16) and mechanic's hands (3/16). Regarding the most frequently antibodies were ANA (100%), anti-RO (41.7%), anti-citrullinated protein antibodies (30%) and rheumatoid factor (RF) (28.6%).

In patients with IPAF, as in the CTD group, the most observed clinical criterion was RP (5/17), followed by arthritis (1/17) and mechanic's hands (1/17). Among the serological criteria the most common antibodies were ANA (100%), followed by anti-RO (33.3%), anti-RNA synthetase (28.6%) and RF (22.2%).

Regarding the radiological pattern, in both groups the most frequent was nonspecific interstitial pneumonia, followed by the indeterminate pattern and usual interstitial pneumonia (UIP) in third place. There were no significant differences by gender and age, between the group of CTD and IPAF, observing in both groups a predominance of women with a similar mean age, being the upper smoking habit in the IPAF group (70.6% vs 31.5%, $p = 0.02$). Regarding the treatment used, the use of immunosuppressants (IS) was more frequent in CTD group (56.3% vs 11.8%, $p = 0.007$).

Conclusion: The clinical-serological and radiological characteristics were similar among patients with IPAF and CTD, which supports the notion of a similar pathophysiology in both groups. In our cohort patients with CTD received IS more frequently than IPAF group, however, future work would be necessary to assess whether the response to treatment is similar in these populations and if IS can benefit patients with IPAF to long term. In addition, it could be useful to include the UIP pattern within the IPAF classification criteria, not currently included, since it is the third most frequent radiological pattern.

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SAT0530

DESCRIPTION AND OUTCOMES OF A SERIES OF 21 PATIENTS WITH INTERSTITIAL PNEUMONIA WITH AUTOIMMUNE FEATURES IN A MULTIDISCIPLINARY UNIT.

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Background: A significant proportion of patients with idiopathic interstitial pneumonia shows autoimmune features, but do not meet the criteria to be classified as a systemic autoimmune disease. In 2015, a joint working group of the European Respiratory Society (ERS) and the American Thoracic Society (ATS) proposed classification criteria for the category called interstitial pneumonia with autoimmune features (IPAF).

Objectives: To evaluate the clinical, serological and morphological characteristics of a series of 21 patients with IPAF as well as the treatment that was applied and the evolution they presented.

Methods: A retrospective cohort study was conducted. The patients who met ERS/ATC IPAF classification criteria in the period from 2012 to 2019 were collected from our interstitial lung disease (ILD) database, including 546 patients. All cases were systematically assessed in a multidisciplinary committee. Clinical, serological, morphological, as well as treatment and outcome variables were collected. A Descriptive analysis is shown.

Results: 21 patients were included in the study, 12 of them (57.1%) women. The mean age at diagnosis was 61.6 years (SD 14.0), and the median follow-up time was 2.9 years (IQR 4.9). All showed ILD by HRCT, 10 (47.6%) patients had autoimmune features at the moment of the diagnosis, and 20 (95.2%) were positive for some auto-antibody. 12 (57.1%) fulfilled two of the three domains, and 9 (42.9%) fulfilled the three IPAF domains. Characteristics are described in Table 1. Lung biopsy was performed in 9 cases (42.9%). The mean FVC at diagnosis was 70.4% (DS 21.0) and DLCO 46.7% (DS 21.5). Regarding the treatment during the disease, mofetil mycophenolate (MMF) was used in 8 (38.1%) patients, cyclophosphamide (CYC) in 3 (14.3%), rituximab (RTX) in 2 (9.5%) and azathioprine in 2 (9.5%). In 1 case (4.8%), CYC and RTX were used in combination. Oral glucocorticoids (GCC) were prescribed in 14 patients (66.7%), 4 of them alone, and 6 cases (28.6%) required high-dose methylprednisolone in 250-1000mg regimens. Antifibrotic drugs (nintedanib or pirfenidone) were used in 5 cases (23.8%) in combination with immunosuppressants and/or GCC. Five patients (23.8%) required home oxygen supplementation. Outcomes to date: 7 (33.3%) patients had readmissions due to ILD acute exacerbation; 2 (9.5%) required lung transplantations, and 3 (14.3%) died during follow-up, 1 (4.8%) due to respiratory causes.

Table 1.

CLINICAL DOMAIN	SEROLOGIC DOMAIN		MORPHOLOGIC DOMAIN		MORPHOLOGY
	n	%	n	%	
					RADIOLOGY
					n %
Raynaud's phenomenon	7	33,3	ANA $\geq$ 1:320 (mottled, diffuse, homogeneous)	12 57,1	NSIP 15 71,4
Inflammatory arthritis polyarticular morning joint stiffness $\geq$ 60 min	4	19	Anti-Ro	11 52,4	UIP 5 23,8
Digital oedema	4	19	ANA nucleolar	3 14,3	LIP 1 4,8
Mechanic hands	1	4,8	ANA centromere	1 4,8	Others 0 0
Distal digital ulceration	1	4,8	Anti-PM-Scl75	4 19	HISTOPATHOLOGY n %
Palmar telangiectasia	1	4,8	Anti-SRP	2 9,5	NSIP 4 19
Gotttron's sign	1	4,8	Anti-PM-Scl100	2 9,5	Interstitial lymphoid aggregates with germinal center 2 9,5
Others	0	0	Rheumatoid factor $\geq$ 2 ULN	2 9,5	Diffuse lymphocyttoplasmic infiltration 1 4,8
			Anti-Jo1	1 4,8	UIP 1 4,8
			PL-7	1 4,8	Others 0 0
			PL-12	1 4,8	MULTICOMPARTMENTAL n %
					AFFECTION
			Anti-Mi2A	1 4,8	Pulmonary vasculopathy 5 23,8
			Anti-CENPA	1 4,8	Pleural effusion 1 4,8
			TIF1-gamma	1 4,8	Intrinsic airways disease 1 4,8
			Others	0 0	Unexplained pericardial effusion 0 0

Conclusion: IPAF was more frequent in women; ANA were the prevalent antibodies; NSIP was the most repeated radiological and pathological pattern; and the combination of GC plus MMF was the most used treatment scheme. Despite our low numbers, acute exacerbations, requirement of lung transplantation and deaths have occurred, which demonstrates the severity of the disease.

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SAT0531

NEXT GENERATION SEQUENCING AND AUTOINFLAMMATORY SYNDROMES: CLINICAL AND GENETICAL CORRELATION ON AN ADULT POPULATION FROM A REFERRAL THIRD-CARE CENTRE

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Background: Systemic autoinflammatory diseases (sAID) are a group of conditions with recurrent episodes of inflammation in absence of infection or autoimmune response. Its pathophysiology mainly lies on mono/poligenic mutations involving genes related to the innate immune system response. Next Generation Sequencing (NGS) platformss have been a big step forward on sAID diagnosis, although a clinical and genetic correlation is still needed.

Objectives: To review the sAID related gene panel variants identified using NGS sAID gene panel on a cohort of adult patients screened for sAID from a referral third-level hospital.

To correlate genetic and clinical findings for sAID related variants identified in order to the clinical suspicion diagnosis of sAID.

Methods: A retrospective review of a cohort of adult ($\geq$ 16 yo) patients with available NGS sAID related gene panel (MiSeq Illumina sequencing platform including intron and exon variants from up to 17 sAID genes, with coverage depth $>$ x100) among 2014 and 2019 was performed.

Demographic, clinical and genetic data were collected in a database.

Genetic variants were classified according to the American College of Medical Genetics/Association for Molecular Pathology classification as benign/likely benign/variable of unknown significance (VUS)/likely pathogenic/pathogenic. In case of polymorphisms or lack of genetic data, the variants were named as unclassified.

A description of the cohort and an analysis of the correlation assessment between clinical data and genetic findings were performed.

Results: 246 out of 299 (82%) patients with NGS sAID gene panel had clinical data available. 170/246 (69%) were adult patients. The medium age was 48 yo, and the M/F ratio was 2.46. 87/170 (51%) adult patients presented 122 variants involving sAID genes (60/87 patients with a single variant). All the variants out of 7 seven were heterozygous variants.

Variants were classified according to ACMG/AMP as follow: pathogenic/probably pathogenic: 22/122 (18%), unknown significance: 74/122 (60.6%), benign/probably benign: 6/122 (4.91%). 20/122 (16.4%) were unclassified variants or polymorphisms.

The most frequent variants identified involved MEFV (54/122), NOD2/CARD15 (18/122) and TNFRSF1A (17/122 including 12 p.Arg121Gln variants) genes.

37/122 (30%) variants correlated with the clinical picture in 33 patients, allowing to confirm the suspected diagnosis. Among the 122 variants, 7 not previously communicated variants were identified.

No somatic variants were found.

Conclusion: NGS sAID related gene panel is a useful tool for sAID diagnosis. In this cohort of 170 adult patients from a referral third-level hospital, genetic tests identified sAID related variants in almost half of them.

20% of patients who underwent genetic NGS sAID related gene panel studies were finally diagnosed with sAID.

The identification of a genetic variant (even pathogenic / likely pathogenic variant) is not diagnostic for sAID if there is not a suggestive clinical picture.

Despite genetic findings, a careful evaluation of clinical – genetic correlation is needed to confirm the suspicion diagnosis, especially for low penetrance variants like TNFRSF1A p.Arg121Gln.

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