Objectives: Aim of the present study was to assess the prevalence of right (RV) or left ventricle (LV) systolic and/or diastolic dysfunction by standard echocardiography and tissue Doppler imaging (TDI).
Methods: Thirty patients with UCTD-risk-SSc (28 female, aged 47±13 years, range 21–70) and 30 age- and sex-matched controls underwent cardiac assessment by standard echocardiography and TDI.
Results: UCTD-risk-SSc patients and controls did not show any difference at standard echocardiographic evaluation. In particular, an inverted E/A ratio was pointed out in 10/30 patients and 7/30 controls (p=ns). TDI showed a mild impairment of LV and RV diastolic (E_m15±4 vs 19±5, $p<0.0001$;E/E_m 6.1±1.7 vs 4.8±1.2, p=0.001; E_t 14±3 vs 16±2, p=0.02; E_t/A_t0.9±0.4 vs 1.3±0.3, $p<0.002$; increased pulmonary artery wedge pressure 9±2 vs 8±1, p=0.001)and systolic function (S_m 13±3 vs 15±2 cm/sec, $p<0.0003$; S_t 14±2 vs 16±3 cm/sec, $p<0.0001$) in UCTD- risk-SSc patients in comparison to controls.
Conclusions: Our study shows that UCTD-risk-SSc patients present a previously unrecognized, mild biventricular systolic and diastolic dysfunction as compared to controls. The pathophysiologic meaning (i.e.intramyocardial artery disease and/or patchy fibrosis) as well the predictive value of developing overt SScin the short time await to be elucidated.
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FRI0379 PREVALENCE OF FAM111 B GENE MUTATIONS IN SYSTEMIC SCLEROSIS

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Background: Systemic sclerosis (SSc) is a prototypic systemic fibrotic disease with unclearly characterized genetic basis. Implicated genes have been associated with autoimmune dysregulation with relatively few variants associated with fibrosis [1]. We have discovered that mutations in *FAM111B* gene cause hereditary fibrosing poikiloderma with tendon contractures, myopathy, and pulmonary fibrosis (POIKTMP)[2], a multisystem fibrotic condition with clinical aspects of SSc [3]. This observation has established *FAM111B* as a candidate gene for SSc.
Objectives:
The objective is to investigate whether *FAM111B* gene mutations are present in SSc patients and further explore relationships between *FAM111B* mutations and clinical expression of SSc.
Methods: Patients with a definite diagnosis of SSc attending the Rheumatology outpatient departments at Groote Schuur Hospital, Cape Town, and Chris Hani Baragwanath Hospital, Johannesburg, were enrolled into the study. Physical examination assessing the extent of disease was done in all patients and the modified Rodnan skin score (mRSS) was used to determine the extent of the skin involvement. Blood samples were collected for DNA extraction and mutation screening using the high-resolution melt technique. Samples with abnormal electropherograms were selected for Sanger sequencing to identify mutations. Public databases were used to verify the frequency of variants in *FAM111B*.
Results: 131 patients were genotyped, 13 men and 118 women, with a mean age of 26.6 years and mean age of symptom onset at 25.3 years. The majority of patients were black (59.5%). 72% of patients had diffuse systemic sclerosis (DSSc) with a median mRSS of 11. Genetic analysis revealed seven rare genetic variants (C832G>A; C855G>T; C917A>G; C937G>A; C988C>T; C995A>C and C1006G>C) in eight patients (five patients from Johannesburg and three patients from Cape Town) [table 1]. These variants were missense mutations of unknown significance with a minor allele frequency <0.01. No *FAM111B* mutations that cause POIKMT were found in patients with SSc.
Conclusions: Rare genetic variants of unknown significance (GVUS) in *FAM111B* gene were found in patients with SSc. It is possible that the GVUS may modify the function of *FAM111B*, and influence the pathogenesis of SSc or are rare polymorphisms with no functional impact.
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FRI0380 PATIENT PREFERENCES AND DIFFICULTIES CONCERNING THE HOME TREATMENT OPTIONS IN SYSTEMIC SCLEROSIS (SSC)

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Background: SSc patients suffer from Raynaud's phenomenon, hand skin hardening and scarring, digital ulcers, esophagus/gastric dysmotility, dysphagia and mucosal dryness. These symptoms significantly reduce patient's life autonomy and impair the capacity to handle their therapy usually made by different modalities of administration: oral, topical, inhalation, intramuscular, subcutaneous, intravenous, and rectal.
Objectives: to investigate the methods of administration used by SSc patients, their preferred methods, their compliance to pharmacological therapy, and the difficulties that are encountered by the patients during pharmacotherapy.
Methods: 2 questionnaires were prepared on an ad hoc basis. The first given to patients, the second to be filled out by the patient's physician. The first questionnaire comprised 25 questions to investigate the problems encountered by patients when taking their medications by correlating these with two validated activities indices: SHAQ-DI (activities of daily living) and the COCHIN scale (hand activities). The second questionnaire collected demographic and important SSc-related clinical information (eg.age, sex, disease duration, symptoms). 80 Ssc patients completed the questionnaires, maintaining anonymity. A "difficulty index" was also filled out, where: 0 = no difficulty to 4 = impossible to use or too difficult to use.
Results:

Table 1. Patient use of pharmacological therapies

	% Using	% Users preferring this method	Difficult index†	Comment
Oral	100	91,25	0,83	Most preferred
Eye drops	52,5	2,5	1,07	Least preferred
Topicals	33,75	3,75	0,77	Cannot open the top (safety lock)
Injections	28,25	2,5	2,17	Cannot apply to skin
Inhaler	16,25	0	1,07	Cannot spray
Vaginal/rectal	6,25	0	1,03	

Table 2. Most frequent problems encountered

	% Using	Most frequent problems	Users have these problems	Difficult index†
Oral	100	Blister: pushing out pills	73,75	2,22
Eye drops	52,5	Open thin stoppers	78,75	1,51
Topicals	33,75	Cannot open the top (safety lock)	70,5	1,4
Inhaler	16,25	Cannot spray/push the inhaler	85	1,71

†4 = most difficult.

In particular, the dimension of the pills is problematic, and has identified a paradoxical situation because large pills are difficult to swallow (41,25% patients who use pills) but at the same time small pills are difficult to pinch with the fingers (62,25% patients who use pills).
Conclusions: SSc patients experience significant problems in maintaining adherence to treatments due to difficulties in the use of blisters and bottles with children proof stoppers. Pills still remain the most preferred method of treatment. In conclusion, patients unanimously wished to avoid the use of blisters, defintively preferring bottles without the children proof stopper to make the treatment easier and provide a more independent life.
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FRI0381 INCREASED RISK OF OSTEOPOROTIC FRACTURES IN ADULT PATIENTS WITH POLYMYOSITIS AND DERMATOMYOSITIS: A NATIONWIDE POPULATION-BASED COHORT STUDY

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Background: Patients with polymyositis and dermatomyositis (PM/DM) are characterized by chronic muscle weakness due to autoimmune-mediated myositis and are usually treated with corticosteroids initially. PM/DM patients prone to develop osteoporosis and subsequent fractures but are rarely investigated.
Objectives: To explore the incidence rate (IR) and risk factors of osteoporotic fractures (OFs) among adult PM/DM patients.
Methods: We conducted a cohort study by utilizing the Taiwan National Health Insurance database. PM/DM patients and respective age- and gender-matched cohort without PM/DM were enrolled. The primary endpoint was the initial event of OFs. We used the Cox proportional hazard model to study the risk factors of OFs in the PM/DM cohort.
Results: Among 2391 PM/DM patients (67.8% female, mean age: 49.5 years) followed for a mean (SD) of 6.1 (5.0) years, 116 developed vertebral fractures, 32 had hip fractures, and 14 experienced radius fractures (IR: 8.18, 2.20, and 0.96 per 1000 person-years, respectively, Table 1). Compared with the matched cohort, the PM/DM patients had higher IR ratios (IRRs) (95% CIs) of OFs at all age groups at enrollment: 3.27 (2.19 to 4.81, $p<0.0001$) for people <50 years and 2.29 (1.85 to 2.82, $p<0.0001$) for those ≥50 years. The IRRs were 2.39 (1.92