MSR Malaysian Society of Rheumatology
23RD MSR - SSR 2025 Workshop in Rheumatology
Singapore Society of Rheumatology

# Young Girl With Tight Skin; A Case Report of LMNA-associated Familiar Partial Lipodystrophy

Farah Nadiah Sulaiman[1], Azrianna Nurfizan Azmi[1], Siti Sanaa Wan Azman[2], Masliza Hanuni Mohd Ali[2], Siti Mariam Ab Rahim[1]
[1]*Rheumatology Unit, Department of Medicine, Hospital Sultanah Nur Zahirah, Terengganu, Malaysia*
[2]*Endorinology Unit, Department of Medicine, Hospital Sultanah Nur Zahirah, Terengganu, Malaysia*

## Introduction

Familial partial lipodystrophy (FPLD) is a rare condition characterized by abnormal distribution of adipose tissue results in its loss from the arms, legs, and hips, but builds up around the face, neck, and abdomen. At least six forms of FPLD are recognized, distinguished by their genetic cause. The most common form is type 2 which is caused by mutations in the LMNA gene. We herein report a case of a patient initially treated as systemic sclerosis but revised to LMNA-associated FPLD.

## Case report

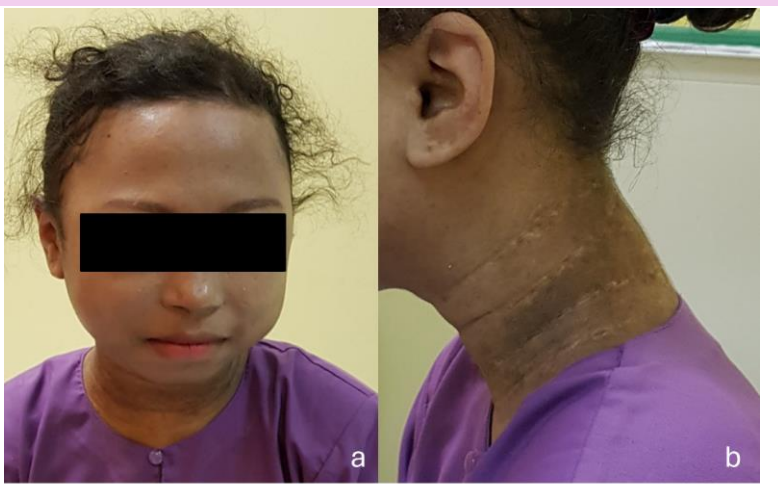
A 23-year-old Malay female was referred to Rheumatology clinic when she was 13 years old for extensive telangiectasia. Clinically, she had skin tightness over bilateral upper and lower limbs, extensive telangiectasia over nails and lower limbs, and calcinosis. There were no features of other connective tissue diseases or complications such as interstitial lung disease or pulmonary hypertension. Skin biopsy showed collagen deposition while anti-nuclear antibody and anti-extractable nuclear antibody were negative. Diagnosis of systemic sclerosis was made thus methotrexate was initiated.

She defaulted follow-up for 2 years and when she presented again, she was diagnosed with diabetes mellitus with HbA1c of 11.1% and negative autoantibodies. She also had unreadable hypertriglyceridemia complicated with admission for severe pancreatitis and lipemia retinalis. Other insulin resistance and metabolic complications were metabolic dysfunction-associated steatotic liver disease, nephropathy, oligomenorrhoea and juvenile osteoporosis. She is resistant to treatment where she requires a high dose of insulin and three types of anti-lipids.

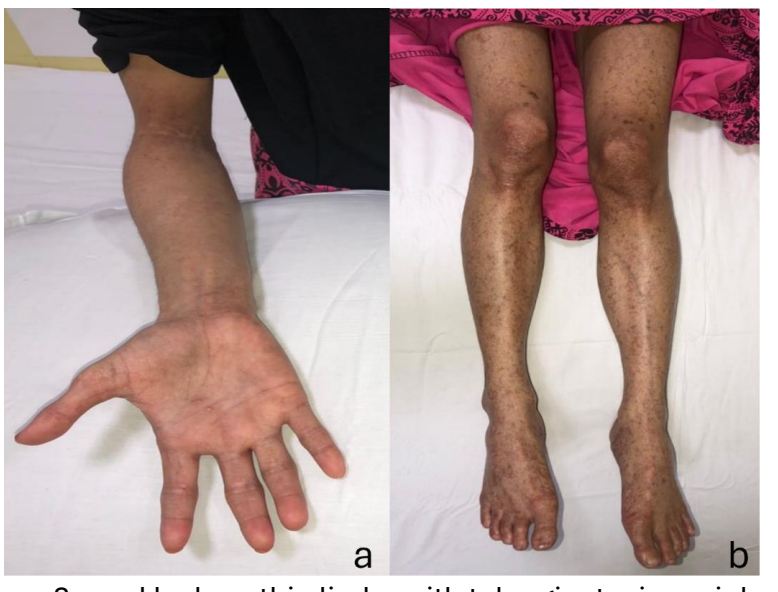
Due to worsening telangiectasia and calcinosis, she was started on prednisolone and methotrexate optimized to 15mg weekly. Colchicine and alendronate were added for persistent calcinosis cutis. Despite multiple medications, her symptoms were not improving.

Additional consult was obtained when she was 20 years old and noticed that she has pterygoid features (small chin, rounded face, prominent eyes) and neck fullness [picture 1a]. She also has acanthosis nigricans [picture 1b], and the previously perceived calcinosis is actually lipid deposits by ultrasound. She also has thin limbs with lower limb telangiectasia [picture 2] which distribution is atypical for systemic sclerosis.

Genetic analysis confirms heterozygous variants of uncertain significance in the LMNA gene suggestive of LMNA-related disorder. There were no other family members with similar features. Her immunosuppressant dose was subsequently tapered off and now planned for leptin replacement therapy.




Picture 1a shows pterygoid features (small chin, round face) while picture 1b shows acanthosis nigricans and fullness over the neck.




Picture 2a and b show thin limbs with telangiectasia mainly at lower limbs and nails. The skin appears tight and shiny.

## Conclusion

FPLD albeit being very rare is an important consideration in a patient presented with atypical features of systemic sclerosis.