



### Lumps and Bumps

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A 19-year-old male born out of third-degree consanguinity presented with insidious onset toe walking and difficulty in getting up from squatting position, thinning of both calves, difficulty in getting up from the chair, not able to walk or run fast, buckling of knees and ankle twisting.

Diamond in Quadriceps sign, Biceps lump, Calf wasting, Extensor Digitorum Brevis hypertrophy and plantar flexion contractures of the ankle all pointed to Dysferlinopathy. (1)

CPK was 22,896 (mcg/L). Genetic testing revealed DYSF+, location Intron49, homozygous, Autosomal Recessive-pathogenic-(LGMD 2, Miyoshi muscular dystrophy -1, Distal Myopathy with anterior tibial onset)



Diamond on Quadriceps sign



Biceps Lump sign

**References**

1. Khadilkar SV, Patel BA, Lalkaka JA. Making sense of the clinical spectrum of limb girdle muscular dystrophies. Pract Neurol. 2018 Jun;18(3):201–10.