

Climb 6-hills in a coat with Popeye, you shall find facioscapulohumeral dystrophy - Going from phenotype to genotype

Suhas M^{1,*}, Sanket Patil¹, and Shivani Nayak²

¹Department of Neurology, Bangalore Medical College and Research Institute, Krishna Rajendra Rd., Bengaluru-560002, Karnataka

²Fellow in Phaco-Refractive Surgery, Lions Super Speciality Eye Hospital, Bengaluru-560002, Karnataka

Abstract

Facioscapulohumeral dystrophy (FSHD), is a rare muscle disease in clinical practice. Identifying the condition with its classic clinical signs helps in better managing patients early. This report is to shed light upon this condition and signs associated with it. We also try to elaborate a few genetic aspects and their pathogenetic mechanisms in the review.

Keywords: Polyhill sign; Popeye sign; coats disease; facioscapulohumeral dystrophy

***Corresponding author:** Dr. Suhas M, Assistant Professor in the Department of Neurology, Bangalore Medical College and Research Institute, Krishna Rajendra Rd., Bengaluru-560002, Karnataka. Email: suhas.murali@gmail.com

Received 8 July 2019; Revised 29 August 2019; Accepted 9 September 2019; Published 23 September 2019

Citation: Suhas M, Patil S, Nayak S. Climb 6-hills in a coat with Popeye, you shall find facioscapulohumeral dystrophy - Going from phenotype to genotype. J Med Sci Res. 2019; 7(4):120-122. DOI: <http://dx.doi.org/10.17727/JMSR.2019/7-21>

Copyright: © 2019 Suhas M et al. Published by KIMS Foundation and Research Center. This is an open-access article distributed under the terms of the Creative Commons Attribution License, which permits unrestricted use, distribution, and reproduction in any medium, provided the original author and source are credited.

Case report

A 27-year-old gentleman presented with a chronic history of weakness and thinning of both upper limbs and difficulty in blowing a whistle, sucking from a straw. There was no significant family history. There was no history of weakness of the lower limbs or involvement of any other part of the neuraxis. On examination he was found to have a classical pattern of weakness involving the trapezius, deltoid, biceps bilaterally but preserved forearm muscles. There was evidence of winging of scapula too. With a pattern diagnosis of facioscapulohumeral dystrophy (FSHD) certain we looked for interesting clinical signs in this patient described below:

Poly-Hill sign

This is due to the selective involvement and sparing of specific muscles all along the shoulder girdle and arm as described below [1]. It consists of a total of 6-Hills (Figure 1).

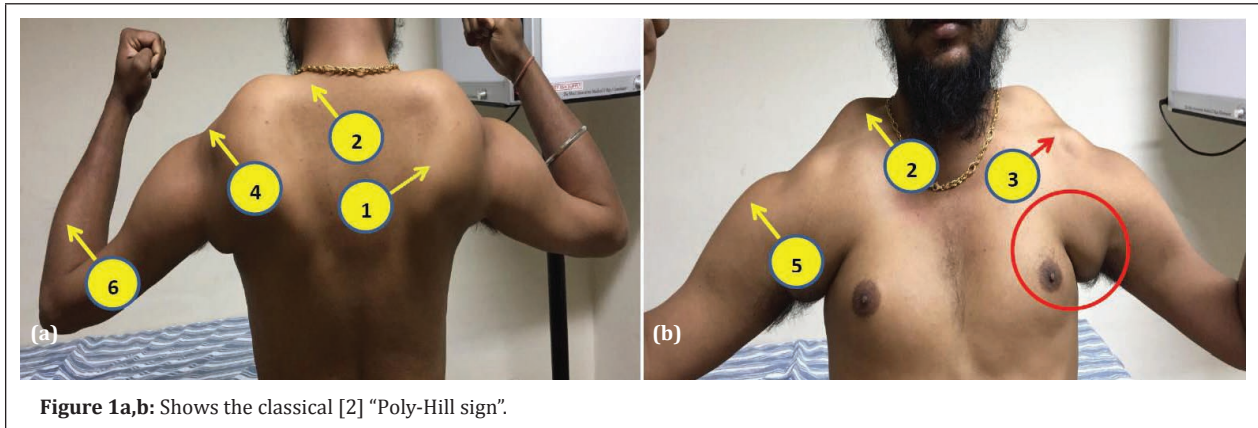


Figure 1a,b: Shows the classical [2] "Poly-Hill sign".

Poly-Hill sign due to:

1. Infraspinal muscle hypertrophy, 2. Upward displacement of the superior scapular angle, 3. Abnormally prominent acromio-clavicular joint (seen from the front in our patient) - Due to the wasting of the trapezius muscle, 4. Partially preserved deltoid muscle - Preserved infero-lateral part due to wasting of the rest of the muscle, 5. Partially preserved biceps brachii muscle - Generally atrophied but some fibers in upper and mid third of the muscle give the appearance of a bump in the muscle, 6. Preserved forearm muscles - mainly the brachioradialis.

In addition to the above there is also "Scapular Winging" due to weakness of Serrati.

Popeye sign [3]:

Interesting history to this sign: The "Popeye sign" (Figure 2) is named after the beloved cartoon character, created by Elzie Segar in the year 1929. It is believed that Segar's hometown was Chester, Illinois, USA. Here in this town there was a gentleman by name "Frank Rocky Fiegel" who is believed to be the actual inspiration behind the character "Popeye" (Figure 3).

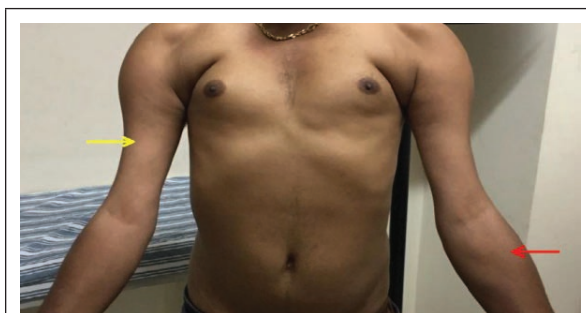


Figure 2: Shows the "Popeye sign" - due to wasting of the biceps brachii (yellow arrow) but spared forearm muscles (red arrow).

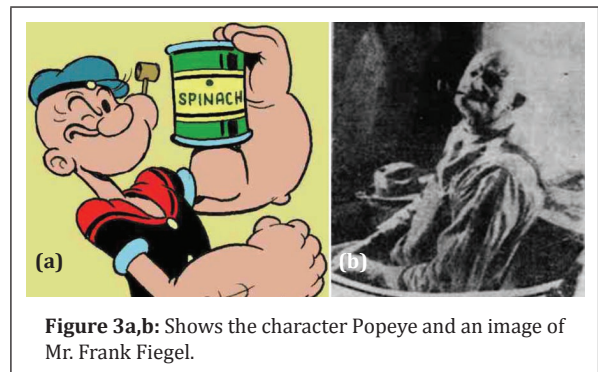


Figure 3a,b: Shows the character Popeye and an image of Mr. Frank Fiegel.

It is interesting to note that the character Popeye, fits the clinical phenotypic description of a case of FSHD. He has classical facial features, wasted biceps and arm muscles and preserved forearm compartment, as shown in figure 4.

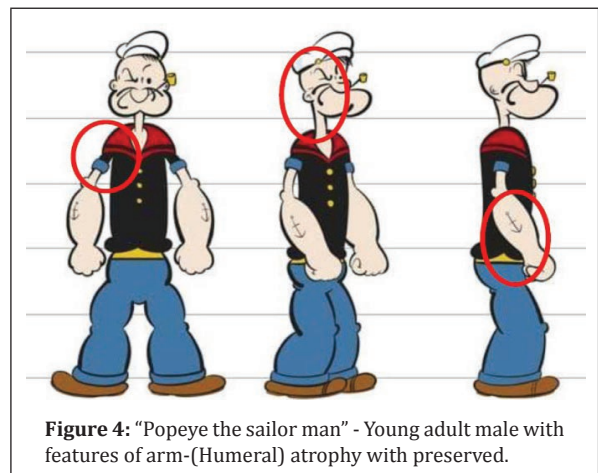


Figure 4: "Popeye the sailor man" - Young adult male with features of arm-(Humeral) atrophy with preserved.

In addition, to these there was evidence of Beever's sign, which is due to asymmetric weakness of the truncal muscles. There is weakness of the rectus abdominis - lower part due to which the umbilicus gets pulled up.

Literature review of FSHD

Pattern recognition approach to muscle disease is the most important clinical skill needed while evaluating a patient with muscle disease. Facio-Scapulo-Humeral- Dystrophy is one such prototype patterned muscle disease [4]. It is an extremely rare condition with some studies estimating its incidence to be about 0.38-0.7/1,00,000 person years [5]. Some studies peg this figure close to 5/1,00,000. It's a congenital disorder which has 2 genetic variants, but both do not differ much phenotypically. The first variant (FSHD-1) is caused due to mutation in the D4Z4 gene in the long arm of Chromosome-4. This variant is by far the most common one encountered, in approximately 95% of FSHD patients. The etiological factors FSHD-2 however, has not been clearly understood.

Phenotypic features of FSHD

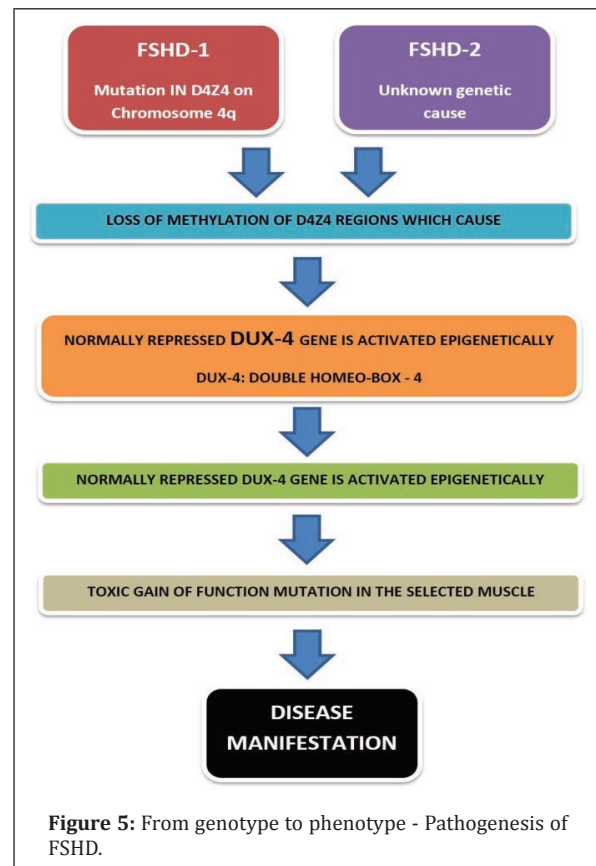
This condition is characterized by Muscular involvement (already described above), extra-muscular involvement in the form of respiratory, cardiac, ophthalmic and ENT abnormalities (Table1).

Table 1: Extra muscular features

<i>Pulmonary</i>	<i>Cardiac</i>	<i>Ophthalmic & ENT</i>
1. Respiratory muscle weakness	1. Dilated cardio myopathy	1. COATS disease - Peripheral retinal telangiectasia
2. Respiratory failure	2. Supra ventricular arrhythmias	2. Hearing loss
3. Kyphoscoliosis	3. Right bundle branch block	

Pathogenesis of FSHD [6]

Follow Up: After doing the literature search we got the genetic analysis done of our patient and he was found to have FSHD-1 genetic variant with a mutation in the D4Z4 region with expression of 8 repeats. It is worth to note that fewer than 11 repeats brings about the phenotypic manifestation of Type-1 FSHD (Figure 5). Also upon follow up for looking at extra-muscular features we also found that our patient had features of COATS disease which he was asymptomatic for. Unfortunately we were not able to get a fundus photograph of the same.



Prognostication

A younger age at presentation, wheelchair bound at an early age, fewer than 10 D4Z4 repeats, early pulmonary involvement are some of the poor prognostic factors in this condition.

Conflicts of interest

Authors declare no conflicts of interest.

References

- [1] Statland JM, Tawil R. Facioscapulohumeral Muscular Dystrophy. Continuum. Minneap. Minn. 2016; 22(6):1916–1931.
- [2] Pradhan S. Poly-hill sign in facioscapulohumeral dystrophy. Muscle Nerve. 2002; 25(5):754–765.
- [3] Cumming WJK. Facioscapulohumeral syndrome. In: Lane RJM, editors. Handbook of muscle disease. Marcel Dekker: New York, 1996; pp. 275–286.
- [4] Pradhan S. An extra hill in a poly-hill sign in a patient with facioscapulohumeral dystrophy. Neurol India. 2007; 55(4):436–447.
- [5] Kurland LT. Descriptive epidemiology of selected neurologic and myopathic disorders with particular reference to a survey in Rochester, Minnesota. J Chronic Dis 1958; 8(4):378–418.
- [6] Lemmers RJ, van d, Klooster R, Sacconi S, Camano P, et al. A unifying genetic model for facioscapulohumeral muscular dystrophy. Science 2010; 329 (5999):1650–1653.