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DERMATOGLYPHIC TRAITS IN CALPAINOPATHY: A CASE REPORT

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ABSTRACT

CASE REPORT

Calpainopathy is a Limb-Girdle Muscular Dystrophy due to calpain deficiency caused by genetic mutations in CAPN 3 gene. It results in progressive, symmetrical weakness of proximal muscles. Other clinical features include tiptoe walking, difficulty in running, scapular winging, waddling gait, scoliosis, calf hypertrophy, and Achilles tendon tightness. Dermatoglyphics or the study of skin patterns in hands and feet can be used to predict the diseases caused by genetic factors, due to the common ectodermal origin of the skin and the nervous system. This study attempts to observe any characteristic dermatoglyphic findings on the hands of a 24-year-old female with Calpainopathy. An electronic flatbed scanner was used to obtain the finger and palm prints which were analyzed using the classification given by Cummins and Mid low. It was observed that ulnar loop patterns were the dominant patterns found with the a-b ridge count from 39 to 41 in both hands. It can be concluded that there is a need for large sample studies to generalize the finding to this population.

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INTRODUCTION

Calpainopathy is described as the autosomal recessive Limb-Girdle Muscular Dystrophy (LGMD) which leads to calpain deficiency caused by mutations in CAPN 3 gene encoding calpain 3 protein. The age of onset is usually in the second decade of life and both sexes are affected. [1]

Calpainopathy is distinguished by the progressive and symmetric weakness of limb-girdle muscles situated proximally. Both intrafamilial and interfamilial phenotype variability, ranging from mild to severe can be seen. Three types of phenotypes have been determined based on the muscle weakness and age of onset: Pelvifemoral limb-girdle muscular dystrophy (LGMD) phenotype,

Scapulohumeral LGMD phenotype, and HyperCKemia. [2]

The clinical features generally observed in calpainopathy are, tiptoe walking, disturbances in running, scapular winging, mild hyperlordosis, waddling gait, laxity of the abdominal muscles, symmetric weakness of proximal muscles in the limbs and trunk. Scoliosis, calf hypertrophy, and Achilles' tendon tightness are also seen. [2]

To improve quality of life and for prolonged survival appropriate physical therapy management is given to each individual. Physical therapy programs should be introduced to promote mobility, maintain joint flexibility, prolong walking and slow the disease progression. Strengthening and gentle low-impact aerobic exercise like swimming, stationary bicycling with supervised sub-maximal effort aids should be done to improve cardiovascular performance, lessen muscle fatigue, and increases muscle efficiency. Canes, walkers, orthotics, and wheelchairs can also be used to compensate for the loss of motor abilities to regain an individual's independence. To prevent contractures use of knee-ankle-foot orthosis at bedtime is recommended. Special attention has to be paid to the seating met wheelchair to avoid scoliosis**.** [2]

Dermatoglyphics is defined as a study of specialized skin patterns of the inter surfaces face of hands and feet. [3] Three types of ridge system are found on distal phalanges: loop, whorl, and arch which account for 60–65%, 30, –35%, and 5 % of all fingerprints, respectively in normal healthy individuals. [4] These ridge patterns depend pon the cornified layer of epithelium and dermal pattern. There is the proliferation of cells in the lower zone of the epidermis which projects in the dermis regular spaced thickenings and the dermis subsequently projects upwards in dermal papillae. This is followed by the appearance of elevations formed by them on the skin surface which are known as epidermal ridges. [5] This specialized skin pattern is formed by the epidermal ridges which constitute the human fingerprint, form during intrauterine life (between 7 -21 weeks), and mature at about 7 months of fetal development. [6] The ridges morphology is genetically determined but it can be influenced by environmental factors during fetal development such as a viral infection, radiation, or alcohol and drug abuse which can disturb brain development. Once, these ridges are matured, they remain unchanged throughout their lifetime. [5]

Since ridges and brain are derived from the ectoderm, so, it might be the reason to use the unusual dermatoglyphic patterns to characterize disturbances to brain development. These prints act as a guide in certain diseases with a proven genetic basis like breast cancer, schizophrenia, Down's syndrome, Klinefelter's syndrome, Alzheimer's disease. [7]

CASE PRESENTATION

A 224-year-old female presented with insidious onset of proximal weakness in both upper limbs and lower limbs for 5 years. She complained of pain in front of the thigh, clean, and back of arm; difficulty in climbing stairs; getting up from squatting position and raising arms above the head. She gets exhausted after walking for a short distance. She was not confident interacting with people due to her altered walking pattern.

Nerve conduction studies showed normal Motor nerve conduction velocity, Sensory nerve conduction velocity, and F-Waves. EMG findings revealed that MUAPs are small, short, and polyphasic. Muscle fiber recruitment was early and full in all the muscles tested. Later, she underwent muscle biopsy examination from the right quadriceps, which revealed the presence of lobulated fibers and incomplete fascicles with variation in fiber size. Immunoblot assay revealed complete loss of calpain and partial loss of Dysferlin which is suggestive of

Calpainopathy. Neurological examination showed normal higher mental functions, cranial nerves, superficial & deep tendon reflexes as well as sensations.

On observation, she is a mesomorph having winging of scapula, rounded shoulders, anterior tilting of the pelvis. She has atrophy of both shoulders with wasting of both deltoids, thinning of thighs, and pseudohypertrophy of both calves. ROM was complete in case of gravity eliminated plane but unable to lift her hand actively beyond 80^{o} against gravity. MMT revealed a muscle power of 3/5 at both shoulders, 4/5 at both elbows, 5/5 at both wrists, 3/5 at both hip joints, 3/5 at both knees, 5/5 at both ankles. Gower's sign was also found to be positive. A waddling gait was found on examination.

She is currently undergoing physical therapy which includes interventions for maintaining and strengthening muscle strength, maintenance of respiratory function, gait training, posture re-education, and endurance training.

There is extensive literature on the characteristic dermatoglyphic findings in medical conditions having a genetic predisposition. This study is an attempt to find characteristic dermatoglyphic traits in a patient with Limb-Girdle Muscular Dystrophy T-IIA.

An electronic flatbed scanner (Canon Lide 300) was used to capture the handprints. The subject was instructed to wash and dry the hands completely before taking the hand scans. The hand was pressed firmly on the flatbed of the scanner with fingers in an abducted position. The fingertip patterns were analyzed and recorded according to the classification given by Cummins and Mid low. The a-b ridge count of both the hands was recorded as it is the most sensitive quantitative dermatoglyphic parameter which is affected by the genetic changes.

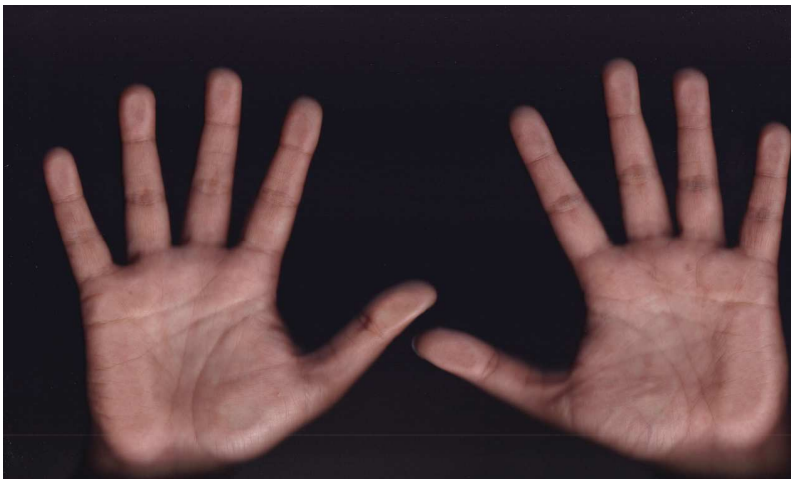

Figure1. Hand scans using electronic Flatbed Scanner

The findings of the dermatoglyphic analysis are summarized in the table below.

Table 1. Descriptive dermatoglyphic findings of both the hands of a subject of Limb-Girdle Muscular Dystrophy

PARAMETERS	**THUMB PATTERN**	**INDEX FINGER PATTERN**	**MIDDLE FINGER PATTERN**	**RING FINGER PATTERN**	**LITTLE FINGER PATTERN**	**a-b RIDGE COUNT**
LEFT HAND	Arch	Ulnar Loop	Ulnar Loop	Ulnar Loop	Ulnar Loop	39
RIGHT HAND	Ulnar Loop	Ulnar Loop	Ulnar Loop	Ulnar Loop	Arch	41

DISCUSSION

These findings are unique in them, however, there is a need for large sample studies revealing the characteristic findings of the dermatoglyphics in patients with muscular dystrophy; the results of which can be generalized on this specific patient population making it easier to screen and detect the risk groups at an early age using dermatoglyphics.

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