International Journal of Science and Research (IJSR)
ISSN: 2319-7064
SJIF (2022): 7.942

Diverse Spectrum of Macrodystrophia Lipomatosa: Case Reports of An Uncommon Condition from Childhood to Adolescence

Dr. Ravi Rajdeo[1], Dr. Miloni Mehta[2], Dr. Divya Agrawal[3], Dr. Sarojini J[4], Dr. Nashreen Fathima[5]

[1]Associate Professor, Government Medical College, Nagpur, India

[2, 3, 4, 5]Junior Residents, Government Medical College, Nagpur, India

Abstract: ***This case study examines the clinical presentation, diagnostic challenges, and management of Macrodystrophia Lipomatosa MDL, an uncommon congenital condition characterized by abnormal growth of adipose tissue. The study involves a series of case reports from childhood to adolescence, highlighting the variability in presentation and progression. MRI imaging was crucial in diagnosis and surgical planning, especially in advanced cases with neurovascular involvement. The findings underscore the importance of early diagnosis and tailored treatment strategies to manage MDLs functional and cosmetic impacts.***

Keywords: Macrodystrophia Lipomatosa, congenital condition, adipose tissue growth, MRI, case report

1. Introduction

This case study examines the clinical presentation, diagnostic challenges, and management of Macrodystrophia Lipomatosa MDL, an uncommon congenital condition characterized by abnormal growth of adipose tissue. The study involves a series of case reports from childhood to adolescence, highlighting the variability in presentation and progression. MRI imaging was crucial in diagnosis and surgical planning, especially in advanced cases with neurovascular involvement. The findings underscore the importance of early diagnosis and tailored treatment strategies to manage MDLs functional and cosmetic impacts.

Purpose Statement

The purpose of this article is to present a series of case reports on Macrodystrophia Lipomatosa, with an emphasis on the diagnostic imaging and clinical management of the condition from childhood to adolescence.

Significance Statement

This study is significant as it provides insights into the varied presentations of Macrodystrophia Lipomatosa, a rare and underreported condition. The findings contribute to better diagnostic accuracy and management strategies, particularly in distinguishing MDL from other similar conditions.

2. Materials and Methods

Study design

It is a observational & descriptive study of MRI extremity in 10 patients presented with enlarged digits.

Inclusion and exclusion criteria

The study included patients referred for MRI of the extremities to assess enlarged digits, excluding those with conditions such as hemangiomatosis, Proteus syndrome, neurofibromatosis, lymphangiomatosis, and Klippel - Trénaunay - Weber syndrome, as well as individuals with metallic implants or claustrophobia.

Representative figure legend

Case 1: A 3 - year - old male child presented with progressive enlargement of the second and third toes of the left foot present since birth.

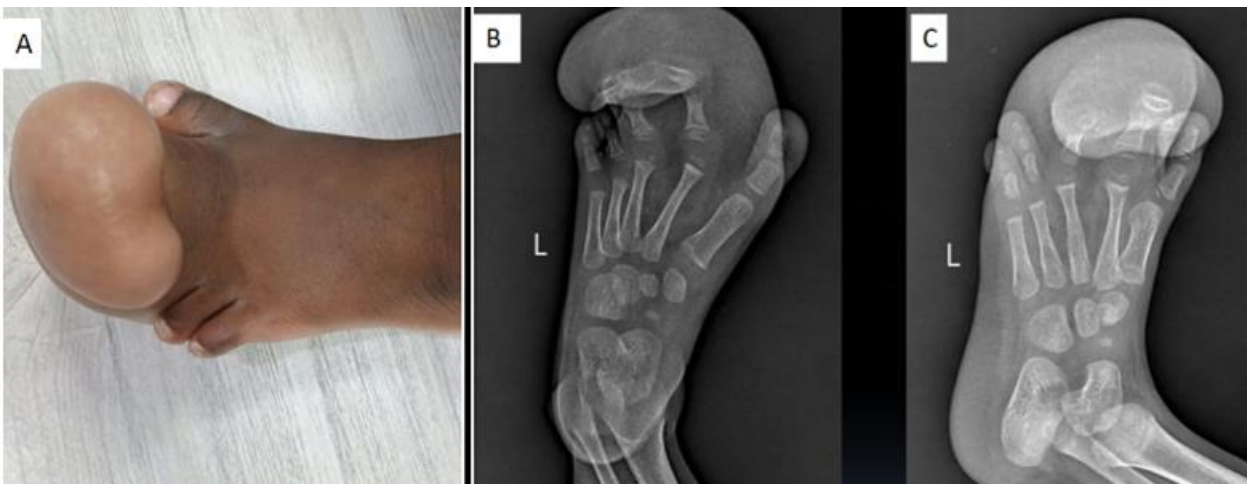

Figure 1: (A) The clinical presentation of the left foot shows distinctive features such as enlargement of the second and third toes with increased soft tissue volume in the forefoot. (B & C) on left foot AP and lateral radiograph, lengthened and

broadened phalanges with endosteal and periosteal bone deposition noted involving 2nd and 3rd toe with overlying soft tissues appears significantly overgrown.

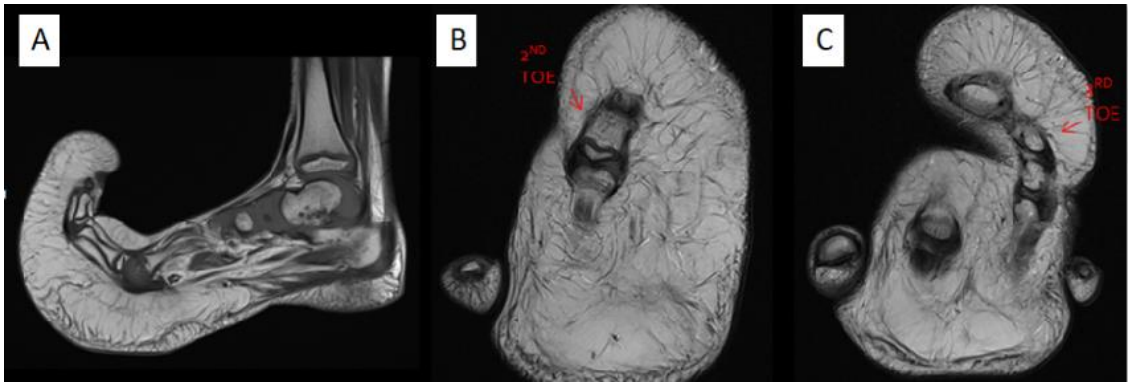

Figure 2: (A), (B) & (C) T1w images showing, diffuse enlargement of left 2nd and 3d toe with markedly increased subcutaneous fatty tissue in the forefoot area and along the plantar fascia with soft tissue syndactyly.

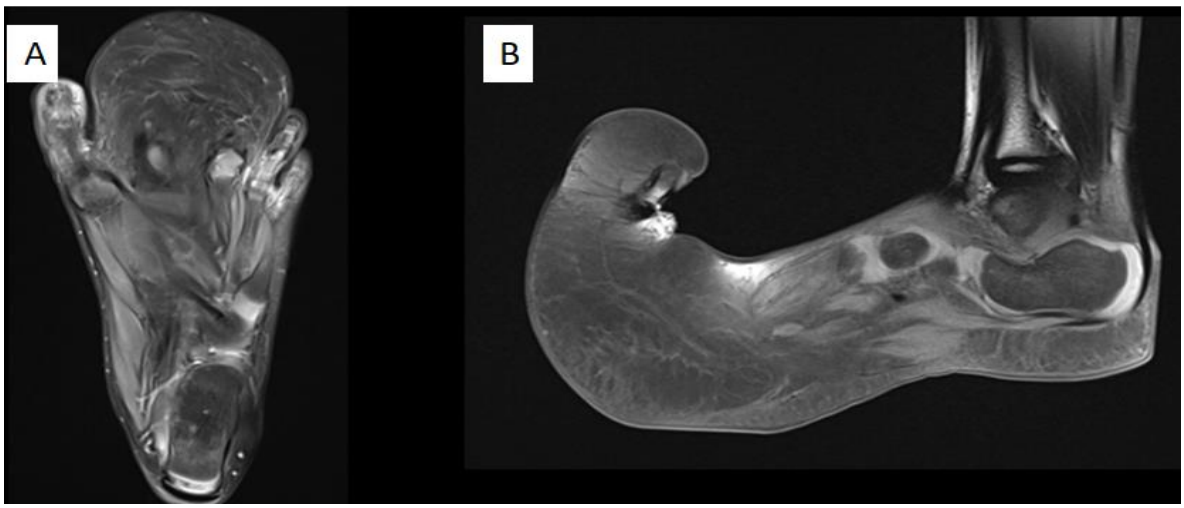

Figure 3: On fat - suppressed sequences (A, B), diffuse signal suppression was observed

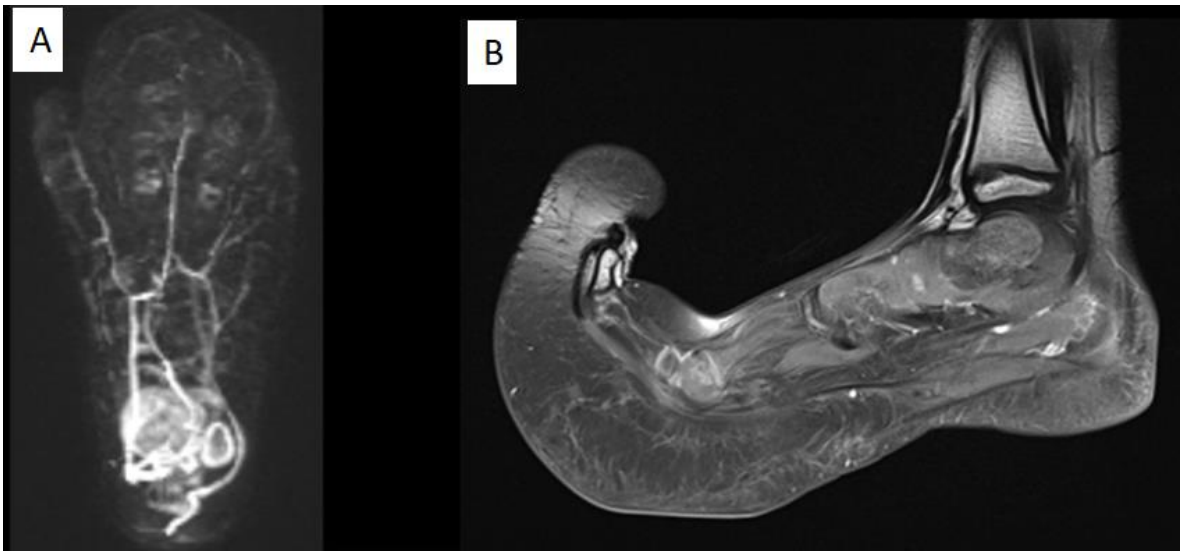

Figure 4: Angiography (A) did not reveal any abnormal vascular channels, and on contrast study (B), there was no abnormal enhancement detected

Case 2: A 2 - year - old child presented with a progressive increase in the size of their thumb over the past year.

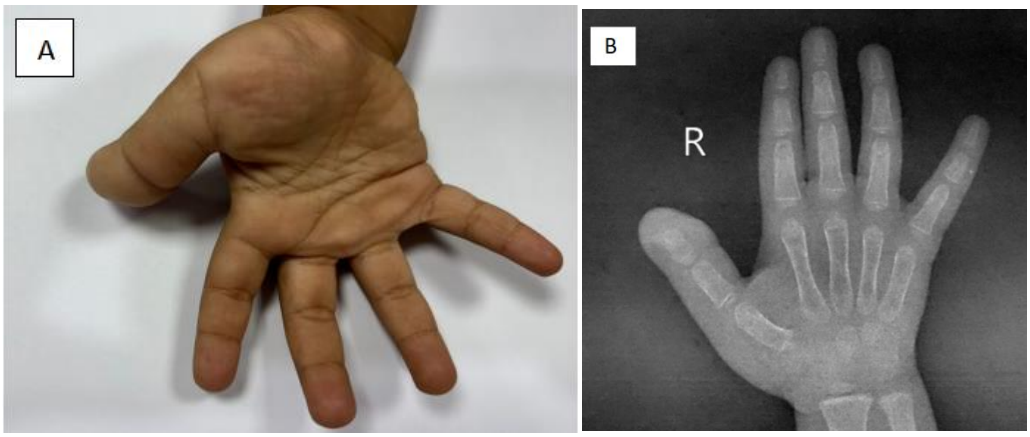

Figure 5: There is mild soft tissue hypertrophy of the thumb with enlargement of the distal phalanx. No associated soft tissue calcification or a focal osseous mass was identified.

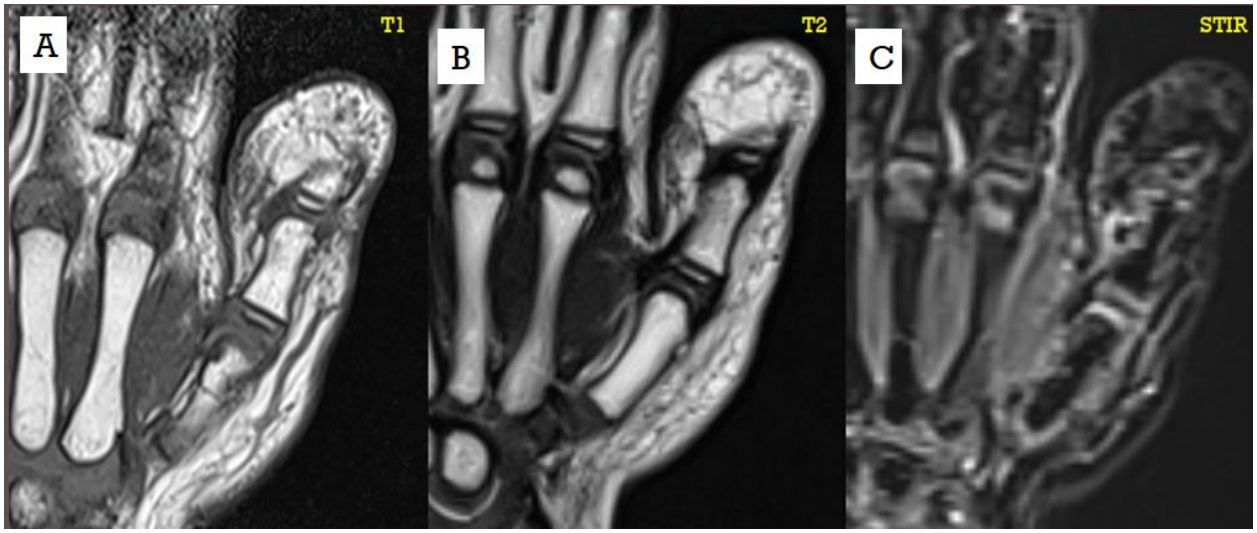

Figure 6: Heterogeneous hyperintensity on T1 (A) and T2 - weighted sequences (B), with signal suppression observed on STIR (short tau inversion recovery) sequences (C).

Case 3: A 16 - year - old female presented with a painless swelling on her right foot that has been present since birth and has gradually increased in size over time.

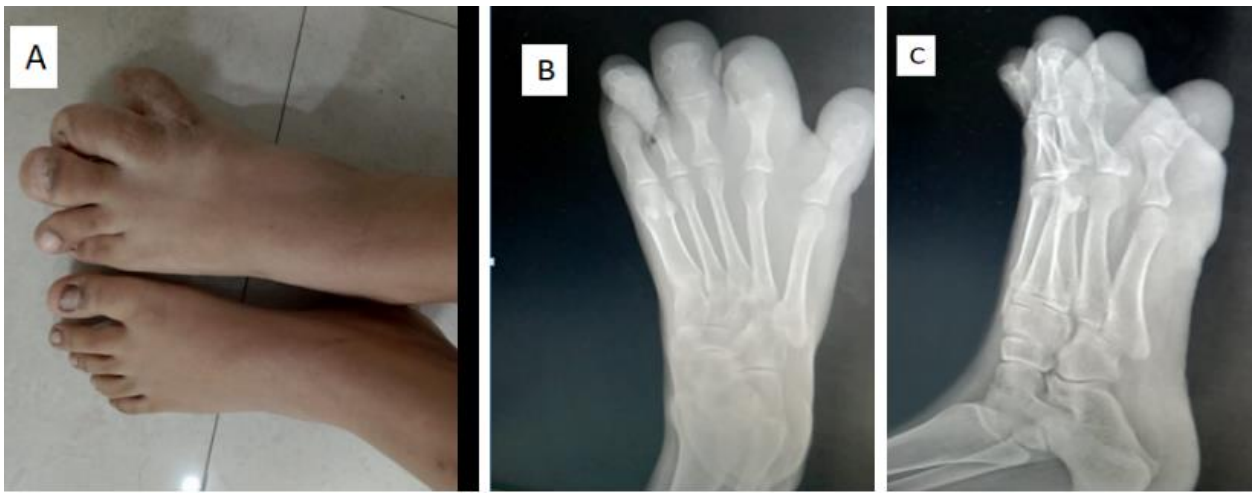

Figure 7: (A) Clinically, the patient presented with diffuse enlargement of the third, fourth, and fifth toes. (B) & (C) AP& lateral radiograph shows osseous hypertrophy of phalanges of the 3rd, 4th and 5th toes

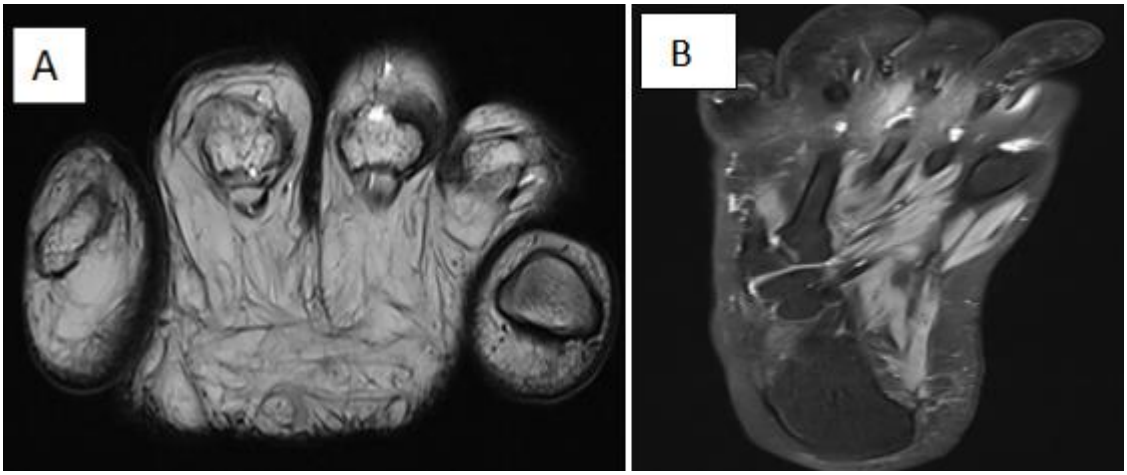

Figure 8: Diffuse enlargement of 3rd, 4th and 5th toes with significantly increased subcutaneous fatty tissue in forefoot region which suprress on fat saturated sequences

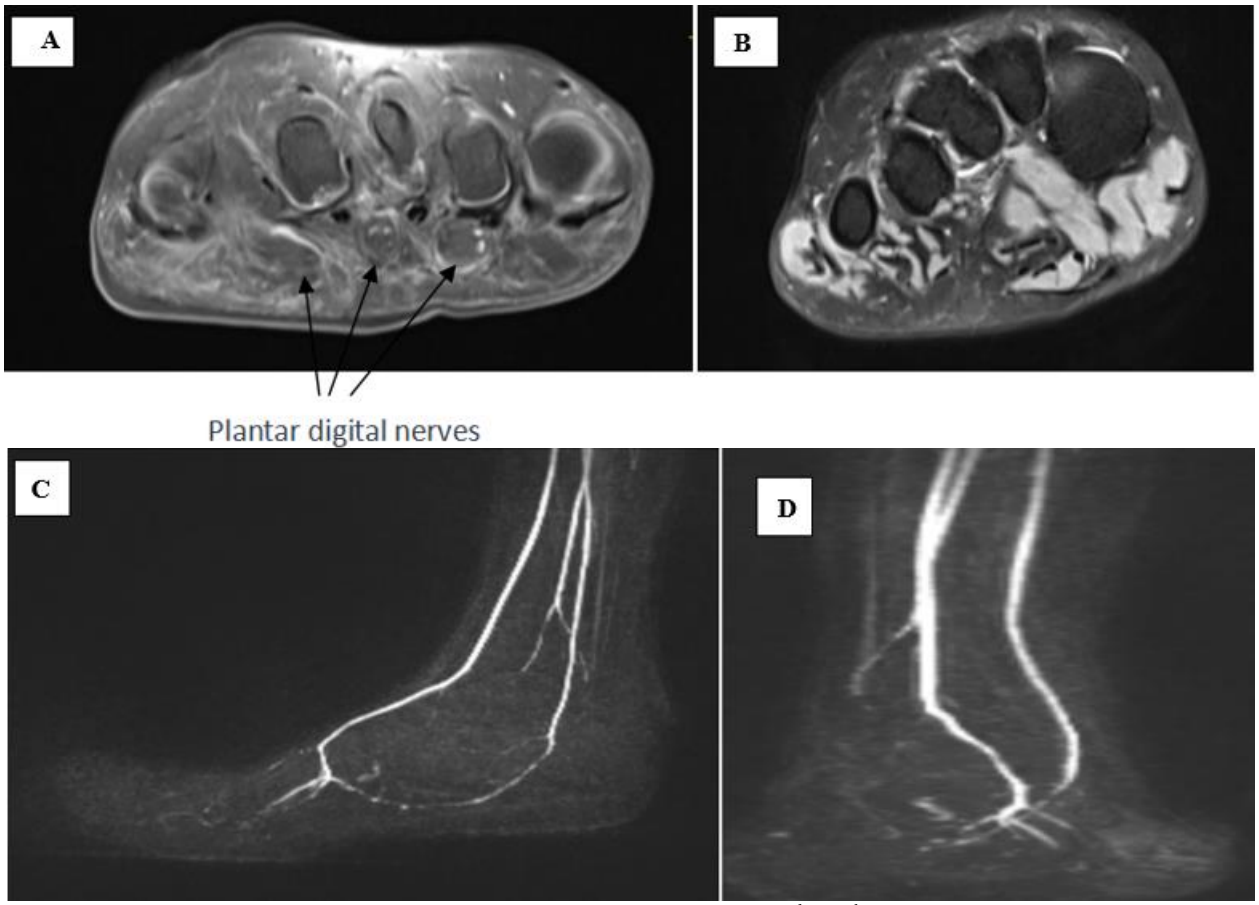

Figure 9: (A) Fusiform enlargement seen involving plantar digital nerves (2nd to 4th webspaces) with fatty proliferation. (B) Severe fatty atrophy of muscles supplied by lateral plantar nerves. (C, D) Significant luminal narrowing of plantar arch and digital branches likley secondary to compression by proliferative fibrofatty tissue.

3. Discussion

Macrodystrophia lipomatosa is a congenital and nonhereditary condition characterized by a progressive persistent proliferation of the mesenchymal components and elements of digits of upper and lower limb, predominantly in median nerve and plantar nerve distribution. Based on the distribution pattern, macrodystrophia lipomatosa is classified into two main types: the "nerve territory oriented type" and the "diffuse or pure lipomatous type" (1)

Macrodystrophia lipomatosa is categorized into two types: static and progressive. The static type is characterized by proportional growth at the affected site, while the progressive type involves disproportionate and ongoing enlargement, distinguishing it as progressive. (3)

Lipofibromatous hamartoma of the nerves often coexists with Macrodystrophia lipomatosa typically affecting the median nerve most frequently. Moreover, secondary degenerative changes and bone deformities are typical characteristics of Macrodystrophia lipomatosa, presenting as new bone growth and bony spurs. (4)

MRI plays a crucial role in evaluating macrodystrophia lipomatosa, particularly for the "diffuse lipomatous" and "mixed pattern" types. MRI is instrumental in assessing the involvement of nerve tissue in macrodystrophia lipomatosa, and it also aids in evaluating the integrity of the neurovascular bundle in conditions associated with syndactly and other related anomalies. (1, 5)

When evaluating patients presenting with hypertrophy and swelling of the hand or foot, differential diagnosis should include several conditions apart from Macrodactly. These include hemangiomatosis, Proteus syndrome, neurofibromatosis, lymphangiomatosis, and Klippel - Trénaunay - Weber syndrome. Each of these conditions presents distinct clinical features that need to be carefully considered to arrive at an accurate diagnosis. (3, 6)

4. Conclusion

Macrodystrophia lipomatosa presents with a broad spectrum of clinical features and progression rates, as demonstrated by the three cases in this study. Early diagnosis and regular monitoring are crucial for managing MDL, given its potential for significant functional impairment and disfigurement. The diversity in presentation—from localized involvement in young children to extensive neurovascular complications in adolescents—highlights the importance of individualized treatment plans. Surgical intervention may be necessary in severe cases to alleviate symptoms and improve quality of life, while milder cases might benefit from conservative management. Further research is needed to better understand the pathophysiology of MDL and to develop more effective therapeutic strategies. Radiographs and MRI, in conjunction with clinical evaluation, are typically adequate for accurate diagnosis. MRI is valuable for assessing the full extent of the condition and for surgical planning purposes.

References

[1] Prabhu C, Madhavi K, Amogh V, Panwala HK, Sathyakumar K. Macrodystrophia Lipomatosa: A Single Large Radiological Study of a Rare Entity. Journal of Clinical Imaging Science [Internet].2019 Feb 27 [cited 2022 Mar 30]; 9: 4. Available from: https: //www.ncbi. nlm. nih. gov/pmc/articles/PMC6702918/
[2] Goldman A, Kaye J. Macrodystrophia lipomatosa: radiographic diagnosis. American Journal of Roentgenology.1977 Jan 1; 128 (1): 101–5.
[3] AlArifi M, Al Essa A, Mashour M, Mohamed Aly A, Tayara B, Al Absi E. Macrodystrophia Lipomatosa of the Finger: A Case Report. Case Reports in Oncology.2019 Jan 21; 12 (1): 63–8.
[4] Inês Da Mata, António Proença Caetano. Macrodystrophia Lipomatosa: A Case Report and Relevant Anatomical Considerations. Journal of the Belgian Society of Radiology.2024 Jan 1; 108 (1).
[5] Dillman JR, Strouse PJ. Macrodystrophia lipomatosa. Pediatric Radiology.2009 Aug 27; 40 (3): 372–2.
[6] Dijendra Nath Biswas, Arkadeep Dhali, Parvin S, Singh A, Gopal Krishna Dhali. Macrodystrophia Lipomatosa: A Rare Cause of Bilateral Lower Limb Gigantism. Curēus.2021 Oct 23.