Open Journal of
Clinical & Medical
Case Reports

Volume 10 (2024)
Issue 11

Short Report ISSN: 2379-1039

Investigation of lipodystrophy in a 3-year-old and 6-month-old child: A case report

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Abstract

This report delineates the case of a 3-year-old and 6-month-old child exhibiting noteworthy weight loss post-vaccination, succeeded by modest adipose tissue gain and subsequent phenotype deterioration following a viral episode. Additionally, the patient presents phlebomegaly, muscular hypertrophy, and irritability linked to hyperphagia. Despite extensive investigation encompassing celiac disease exclusion, the prevailing diagnostic hypothesis is acquired generalized lipodystrophy.

Keywords: Lipodystrophy; Weight loss; Hyperphagia; Muscular hypertrophy; Phenotype.

Abbreviations: APLV: Allergy Protein Milk Cow's; DM1: Type 1 Diabetes Mellitus; HAS: Hypertension Arterial Systemic.

Introduction

Lipodystrophy, a rare condition characterized by selective, localized, partial, or nearly generalized loss of subcutaneous adipose tissue, presents both acquired and congenital forms. Acquired lipodystrophy may manifest in tandem with various conditions, including viral infections, autoimmune diseases, metabolic disorders, and adverse drug reactions. This case report elucidates a child with acquired generalized lipodystrophy whose clinical manifestation and progression posed diagnostic challenges.

Case Presentation

The patient, a 3-year-old and 6-month-old girl, delivered via cesarean section with a birth weight of 3600 g and length of 51 cm, displayed a typical adipose tissue distribution. Despite experiencing cow's milk protein allergy necessitating infant formula due to inadequate breastfeeding, she exhibited appropriate weight, height gain, and neuropsychomotor development for her age. Following poliomyelitis vaccination at one year and three months, she underwent rapid and significant weight loss (approximately 1.7% in a few days), primarily in the limbs, followed by modest weight gain while maintaining reduced adipose tissue in the areas above. However, after a viral episode in October 2023, a deterioration in phenotype ensued, characterized by phlebomegaly, muscular hypertrophy, and irritability associated with hyperphagia (Figure 1). Extensive etiological investigation revealed hypertriglyceridemia, hepatomegaly with hepatic steatosis, altered transaminases, and hyperinsulinemia. Leptin levels were undetectable, Complement 4 (C4) levels below the lower limit of normality, and viral infections such as HIV and hepatitis B and C were ruled out. The patient did not present hematological neoplasms, undergo bone marrow transplantation, or use checkpoint inhibitors.

Furthermore, she exhibited no autoimmune diseases. Genetic testing for candidate lipodystrophy genes yielded no suggestive genes. The parents, although non-consanguineous and devoid of a similar phenotype, had the father diagnosed with type 1 diabetes at age 4.

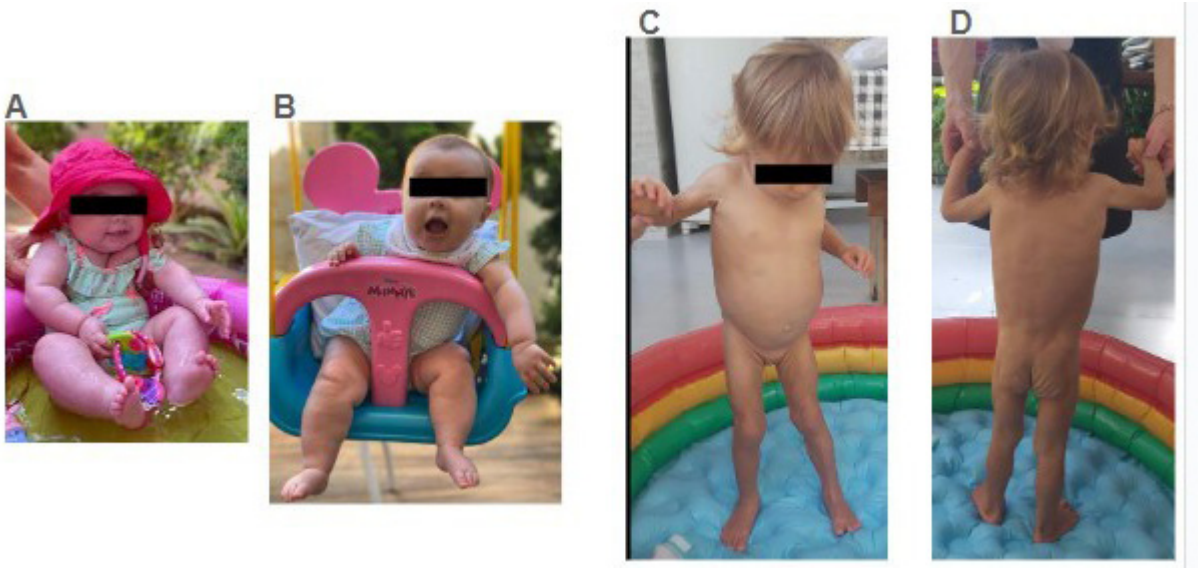

Figure 1: Phenotypic variation. A and B: before one year and three months. **C,D**: after starting the loss of adipose tissue.

Discussion/Conclusion

Acquired generalized lipodystrophy, or Lawrence Syndrome, represents a rare lipodystrophic syndrome characterized by adipose tissue loss, insulin resistance, and an elevated cardiovascular risk. Over 100 cases have been documented, with a female-to-male ratio of 3:1. The clinical presentation mirrors that of Berardinelli-Seip syndrome, suggesting an acquired syndrome. Differential diagnoses include other forms of extreme insulin resistance and various lipodystrophies. Management primarily addresses metabolic

manifestations akin to other insulin resistance syndromes, with treatment modalities such as lifestyle modifications, insulin sensitizers, antihypertensives, and hypertriglyceridemia management. Recombinant human leptin has demonstrated efficacy in some cases. The prognosis remains uncertain, likely influenced by cardiovascular risk and underlying etiology. Lawrence syndrome, or Acquired Generalized Lipodystrophy (AGL), signifies a rare acquired condition characterized by substantial fat loss across multiple body regions, notably the face, arms, and legs. While sharing similarities with other lipodystrophies, the etiology and progression of AGL remain distinct. This case underscores the diagnostic complexity of lipodystrophy in children, emphasizing the necessity for a multidisciplinary approach to ensure optimal growth and development [1-7].

Declarations

Acknowledgements: The authors thank the patient's family for their collaboration and consent to share this case.

Financial support: There was no financial support.

Competing interests: Authors state no conflict of interest relevant to this manuscript.

Availability of data and material: Original data generated and analyzed are available through request to the corresponding authors.

Disclosure statement: All authors have nothing to disclose with respect to this manuscript.

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Manuscript Information: Received: March 12, 2024; Accepted: April 25, 2024; Published: April 30, 2024

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Citation: Guidorizzi NR, Foss-Freitas MC. Investigation of lipodystrophy in a 3-year-old and 6-month-old child: A case report. Open J Clin Med Case Rep. 2024; 2233.