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Life without fat: A case report on familial partial lipodystrophy, dunnigantype in a 10-year old female

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Abstract

Statement of the Problem: Familial partial lipodystrophy of the Dunnigan type is a rare autosomal dominant disorder. It is frequently unrecognized, with no firm diagnostic criteria established. This case report aims to improve the clinical suspicion as well as to discuss the pathophysiology as well as review the current knowledge, trends and issues surrounding the diagnosis and treatment of lipodystrophy.

Methodology & Theoretical Orientation: We examined a suspected case of lipodystrophy in a 10-year old child. Patient underwent a series of diagnostic and laboratory work-up. Genetic analysis was also done. Literature was reviewed regarding the diagnosis and management of lipodystrophy, particularly in the pediatric population.

Findings: This is a case of a 10-year-old Filipino diabetic female with a history of persistently elevated blood glucose. On physical examination, she demonstrated loss of subcutaneous tissue in the extremities and gluteal area, with accumulation of fat on the face, neck and abdominal area. Further work-up revealed hyperinsulinemia with insulin resistance, dyslipidemia and focal fat infiltrated areas in the left liver lobe. Dual-energy x-ray absorptiometry showed a higher percentage of fat tissue in the trunk compared to the extremities. High-resolution chromosomal analysis revealed a normal female chromosome complement in all cells examined.

Conclusion & Significance: Diagnosis of lipodystrophy is primarily based on history, physical examination, body composition and metabolic status. More often than not, it can lead to misdiagnosis with metabolic syndrome, diabetes or Cushing syndrome. This case highlights the importance on the awareness of the triad of lipodystrophy followed by careful clinical assessment of fat distribution. This leads to earlier detection hereby preventing further complications.



Biography:

Rinah Elaisse Dolores has completed her MD-MBA at the age of 27 years from Ateneo School of Medicine and Public Health. She is currently a 1st year pediatric resident at National Children's Hospital in Manila, Philippines.



Speaker Publications:

 Life without fat: A case report on familial partial lipodystrophy, dunnigan-type in a 10-year old female. Publication date April 15, 2020 publication description International Journal of Environmental Research and Public Health.

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