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CASE REPORT

Mauriac syndrome in an adolescent – case report: a rare complication of type 1 diabetes mellitus

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Abstract

Introduction: Mauriac syndrome is a rare complication of type 1 diabetes mellitus associated with poor metabolic control, which is particularly rare since the introduction of intensive insulin therapy. It is characterized by hepatomegaly, dyslipidemia, growth failure, and delayed puberty. **Case report:** We present the case of a 14-year-old male with Mauriac syndrome, with poor metabolic control since his diagnosis of type 1 diabetes mellitus at age two, with further deterioration during adolescence due to insulin omission and consultation absenteeism. Early recognition of this syndrome is crucial, given its reversibility with appropriate glycemic control. **Discussion:** This case underscores the importance of a personalized approach to patient care, highlighting the challenges of therapeutic compliance during adolescence, which often necessitates a multidisciplinary team and a comprehensive understanding of the patient's circumstances.

Keywords: Type 1 diabetes mellitus. Diabetes complications. Adolescent. Hepatomegaly.

Síndrome de Mauriac num adolescente – caso clínico: uma complicação rara da diabetes mellitus tipo 1

Resumo

Introdução: A síndrome de Mauriac é uma complicação da diabetes mellitus tipo 1 com mau controlo metabólico, muito rara desde a introdução da terapêutica intensiva com insulina. Caracteriza-se por hepatomegalia, dislipidemia, atraso do crescimento estaturo-ponderal e atraso pubertário. **Relato de caso:** Descrevemos o caso de um adolescente do sexo masculino de 14 anos com síndrome de Mauriac, com mau controlo metabólico desde o diagnóstico de diabetes mellitus tipo 1 (2 anos), mais relevante desde a entrada na adolescência, por omissão da administração da insulina e absentismo às consultas. O reconhecimento precoce desta entidade é extremamente importante, dada a reversibilidade com o controlo metabólico adequado. **Discussão:** Este caso pretende sublinhar a importância de uma abordagem personalizada a cada doente, uma vez que a adesão à terapêutica pode ser muito desafiante, sobretudo durante a adolescência, por vezes necessitando do trabalho em equipa multidisciplinar e a compreensão dos variados aspetos de cada doente.

Palavras-chave: Diabetes mellitus tipo 1. Complicações de diabetes. Adolescente. Hepatomegalia.

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Keypoints

What is known

- Mauriac syndrome is a complication of type 1 diabetes mellitus with poor metabolic control, that has become a rarity since the introduction of intensive insulin therapy.
- It is characterized by hepatomegaly, dyslipidemia, growth failure, and delayed puberty.
- Early recognition of MS is extremely important, given its reversibility with adequate glycemic control.

What is added

- There are multifaceted challenges in managing type 1 diabetes mellitus during adolescence, particularly the impact of social, emotional, and behavioral factors on therapeutic compliance, requiring the incorporation of psychological and social support into medical care.
- The unique revelation of the patient's competence in managing insulin dosages while caring for his younger sister underscores the complex interplay between family dynamics and self-care in chronic disease management.

Introduction

Type 1 diabetes mellitus (T1DM) is a chronic metabolic disease affecting 0.16% of the Portuguese population as of 2015[1]. Mauriac syndrome (MS), is a complication of T1DM, primarily affecting adolescents with poor glycemic control. It is characterized by hepatomegaly due to hepatic glycogenosis (HG), dyslipidemia, growth failure, and delayed puberty. This syndrome has become rare since the introduction of intensive insulin therapy[2].

The physiopathology of MS is not fully understood and likely results from a combination of factors, including wide fluctuations in plasma glucose levels, periods of hyperglycemia, and hyperinsulinism[2]. Early recognition is essential as it is fully reversible with adequate glycemic control[3-5]. However, achieving therapeutic compliance can be challenging during adolescence[4], often requiring a comprehensive multidisciplinary approach.

Case report

We present the case of a 14-year-old male diagnosed with T1DM at the age of two, from a low socioeconomic background. His family history includes an eight-year-old sister with T1DM and celiac disease. He has exhibited poor metabolic control since diagnosis, which worsened during adolescence, with multiple emergency department admissions due to diabetic ketoacidosis and recurrent hypoglycemic episodes. The patient was on intensive insulin therapy (1.3 units/kg/day). Despite the efforts of a multidisciplinary team and structured admissions to optimize metabolic control and educate the patient and family, a pattern of consultation absenteeism and insulin omission emerged, particularly during the COVID-19 pandemic.

Follow-up revealed suboptimal glycemic control, abdominal distention, and growth retardation. His stature was below the 3rd percentile (Z-score—6.72; growth rate: 2 cm/year) (Fig. 1) and his body mass index was 16.1 kg/m² (percentile 3). Physical examination revealed a protuberant abdomen and hepatomegaly, with no jaundice, splenomegaly, oedema, or ascites. Secondary sexual characters were absent (Tanner stage 1), with a two-year delay in bone age.

Laboratory analysis showed poor metabolic control: glycemia 305 mg/dL, HbA1c 11% (72% above target); dyslipidemia (triglycerides 277 mg/dL; total cholesterol 170 mg/dL; low-density lipoprotein 56 mg/dL), and elevated hepatic enzymes (aspartate aminotransferase 63 U/L; alanine aminotransferase 109 U/L). Hormone levels were pre-pubertal, with suboptimal FSH, LH, and testosterone readings. Insulin Growth Factor-1 (IGF-1) was also below normal (92 ng/mL). Hemogram, coagulation, renal function, iron metabolism, and bilirubin were normal. Microalbuminuria was under 1.10 μg/dL. Viral serologies were negative for hepatitis A, B, C, and HIV. Liver autoantibodies, including anti-mitochondrial, anti-smooth muscle, and anti-liver-kidney microsome antibodies, were negative. Inborn errors of metabolism profiles were normal. Anti-thyroglobulin, anti-transglutaminase, and anti-gliadin antibodies were also negative.

Abdominal ultrasonography confirmed moderate hepatomegaly, with a regular liver contour and homogenous echostructure.

The combination of hepatomegaly, elevated aminotransferases, dyslipidemia, growth failure, and delayed puberty in an adolescent with T1DM and poor metabolic control strongly indicated MS. He was electively admitted for one month to achieve metabolic control. During this period, the multidisciplinary team's efforts, including pharmacological therapy with antidepressants and individual and family therapy sessions, were crucial. Discussions addressing the patient's emotional challenges (emotional turmoil and despair facing his condition), fear of needles, and insulin omission at school facilitated therapeutic compliance. He was discharged with an insulin dose of 1 unit/kg/day, HbA1c 7% and normalization of aminotransferases, although lipid profiles remained abnormal.

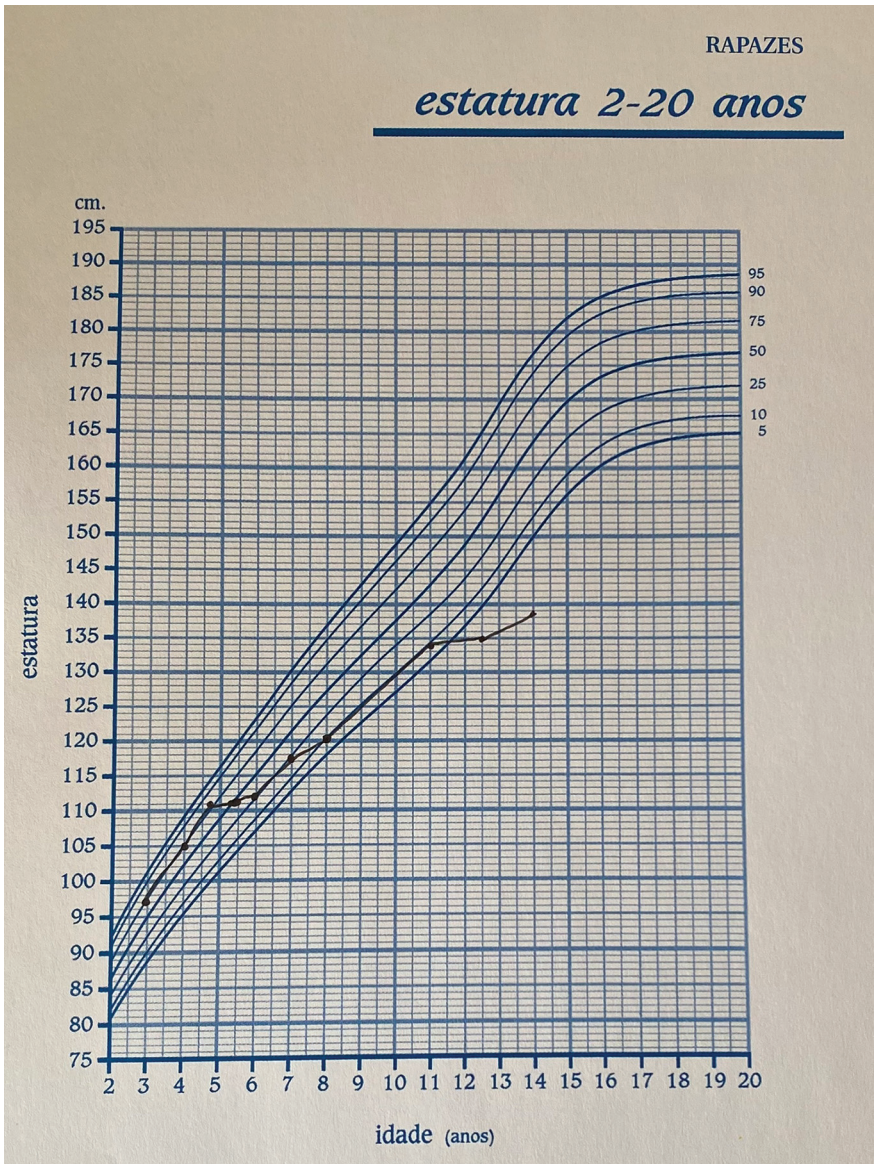

Figure 1. Graph showing the adolescent's evolution of height (cm) over his age (years), revealing the progressive reduction in growth rate.

Six months later, the patient showed positive progress with good metabolic control (HbA1c 7.7%, 82% on target), age-appropriate puberty hormones, normalization of IGF-1, and a growth of 3.2 cm. Statin therapy was initiated for persistent dyslipidemia.

Discussion

In MS, hepatomegaly is caused by hepatic glycogenosis (HG). High glucose levels promote the transport of glucose into hepatocytes and hyperinsulinemia stimulates glycogen synthesis. Episodes of hyperinsulinism, with transient hypoglycemia, stimulate cortisol production, leading to further hepatic deposition of glycogen-causing elevated aminotransferases[5-6].

In Portugal, the exact incidence of Mauriac Syndrome is not well-documented, reflecting its rarity in the modern era of intensive T1DM care. This case underscores the importance of recognizing MS, even in the context of advanced insulin therapies, particularly in adolescents with poor glycemic control. The presence of significant social, emotional, and behavioral factors, such as insulin omission and consultation absenteeism exacerbated by the COVID-19 pandemic, highlights the

multifaceted challenges in managing T1DM during adolescence.

This case contributes to the existing literature by providing a contemporary example of MS in a Portuguese adolescent, emphasizing the ongoing need for vigilance for this condition. It also underscores the importance of a multidisciplinary and personalized approach to T1DM management, particularly during adolescence, a period marked by unique adherence challenges.

The differential diagnosis includes other causes of liver injury, such as infectious, metabolic, obstructive, or autoimmune diseases[5]. One important differential diagnosis is non-alcoholic fatty liver disease (NAFLD), more common in type 2 diabetes mellitus or obese patients[5-8]. The final diagnosis usually involves a liver biopsy[3,5-8]. In our patient, the presence of hepatomegaly, elevated aminotransferases, dyslipidemia, growth failure, delayed puberty in an adolescent with T1DM, and poor metabolic control strongly suggested HG. The normalization of hepatomegaly and liver enzymes after four weeks of good metabolic control corroborates this hypothesis, enabling us to avoid an invasive examination, such as a liver biopsy[3,7-8].

Various mechanisms may contribute to growth retardation in MS, including increased growth hormone (GH), decreased IGF-1, impaired hormone bioactivity, and resistant or defective hormone receptors. Autoimmune conditions, like celiac disease and hypothyroidism, could also cause growth delay in T1DM patients[9]. However, relevant antibodies (anti-thyroglobulin, anti-transglutaminase, and anti-gliadin) were negative in our patient.

Therapeutic adherence is particularly challenging during adolescence, often requiring medical, psychological, and psychiatric interventions[4,9]. This complex landscape requires balancing understanding and acceptance of the condition while fostering patient autonomy. A notable aspect of our patient's journey was his competence in insulin dosage calculation and administration, a skill developed while caring for his younger sister. This dual role paradoxically correlated with proficient metabolic control, highlighting the practical aspects of self-care beyond theoretical understanding.

Improved metabolic control during hospitalization and an enhanced well-being gave the patient the confidence to change. This case underscores the intricate challenges of treatment adherence in chronic adolescent conditions. Multidisciplinary teamwork was crucial, integrating meticulous metabolic control, close surveillance, and fostering condition acceptance and emotional support.

This case highlights the importance of recognizing rare pathologies like Mauriac Syndrome, particularly in the context of treatment adherence challenges. It also emphasizes the necessity of multidisciplinary team interventions in managing T1DM, addressing both the patient and their family as a whole. We believe that the diagnosis of a chronic disease like T1DM should always involve multidisciplinary consultations for optimal glycemic control, complication prevention, and overall well-being and self-care.

Author contributions

B. Moreira, T. Brito, I. Fernandes: Conception and design of the case report; Acquisition of data either from patients, research studies, or literature; Analysis and interpretation of data either from patients, research studies and literature; Drafting the article; Critical review of the article for important intellectual content; Final approval of the version to be published. S. Parente: Final approval of the version to be published; Agreement to be accountable for the accuracy or integrity of the work.

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Conflicts of interest

None.

Ethical considerations

Protection of humans and animals. The authors declare that the procedures followed complied with the ethical standards of the responsible human experimentation committee and adhered to the World Medical Association and the Declaration of Helsinki. The procedures were approved by the institutional Ethics Committee.

Confidentiality, informed consent, and ethical approval. The authors have followed their institution's confidentiality protocols, obtained informed consent from patients, and received approval from the Ethics Committee. The SAGER guidelines were followed according to the nature of the study.

Declaration on the use of artificial intelligence. The authors declare that no generative artificial intelligence was used in the writing of this manuscript.

References

1. Diabetes: Factos e Números – O Ano de 2015 – Relatório Anual do Observatório Nacional da Diabetes. Lisboa: Sociedade Portuguesa de Diabetologia. Available at: https://www.sns.gov.pt/wp-content/uploads/2017/03/OND-2017_Anexo2.pdf

2. Alhajjaj AH, Aljishi FK. *Mauriac Syndrome Still Exists in Poorly Controlled Type 1 Diabetes: A Report of Two Cases and Literature Review*. Cureus. 2021;13(4):e14704. Published 2021 Apr 26. doi:10.7759/cureus.14704
3. Pinto MJF, Melo N, Flores L, Cunha F. *Mauriac Syndrome: A Rare Complication of Type 1 Diabetes Mellitus*. Eur J Case Rep Intern Med. 2018;5(12):000969. Published 2018 Dec 27. doi:10.12890/2018_000969
4. Moreno Tirado A, González Lázaro P, Montero Benítez MZ, Jiménez Torrecilla P. *Hepatopathy of Mauriac syndrome. The importance of therapeutic adherence* [published online ahead of print, 2023 Feb 13]. Rev Esp Enferm Dig. 2023;10.17235/reed.2023.9513/2023. doi:10.17235/reed.2023.9513/2023
5. Patita M, Nunes G, Alves de Matos A, Coelho H, Fonseca C, Fonseca J. *Mauriac Syndrome: A Rare Hepatic Glycogenosis in Poorly Controlled Type 1 Diabetes*. GE Port J Gastroenterol. 2019;26(5):370-374. doi:10.1159/000496094
6. Abu NA, Lim CB, Nor NSM. *Glycogenic hepatopathy in children with poorly controlled type 1 diabetes mellitus*. Clin Pediatr Endocrinol. 2021;30(2):93-97. doi:10.1297/cpe.30.93
7. Lombardo F, Passanisi S, Gasbarro A, Tuccari G, Ieni A, Salzano G. *Hepatomegaly and type 1 diabetes: a clinical case of Mauriac's syndrome*. Ital J Pediatr. 2019;45(1):3. Published 2019 Jan 7. doi:10.1186/s13052-018-0598-2
8. Khoury J, Zohar Y, Shehadeh N, Saadi T. *Glycogenic hepatopathy*. Hepatobiliary Pancreat Dis Int. 2018;17(2):113-118. doi:10.1016/j.hbpd.2018.02.006
9. Bizzarri C, Timpanaro TA, Matteoli MC, Patera IP, Cappa M, Cianfarani S. Growth *Trajectory in Children with Type 1 Diabetes Mellitus: The Impact of Insulin Treatment and Metabolic Control*. Horm Res Paediatr. 2018;89(3):172-177. doi:10.1159/000486698