

31^o CURSO PÓS-GRADUADO ENDOCRINOLOGIA DIABETES E METABOLISMO

POSTERS | MEDICINA GERAL E FAMILIAR

MGF01

INTERVENTION PROJECT: HEALTHY EATING RECOMMENDATIONS FOR CHILDREN WITH OVERWEIGHT/OBESITY - IMPACT ON BMI

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Background/Aims: Healthy eating plays a fundamental role in childhood development. Childhood overweight/obesity constitute a major global public health problem, so educational interventions targeting them and their caregivers may help modify health-related behaviours. The aim of this intervention was to assess the impact of an educational intervention on healthy eating in children with overweight/obesity, by measuring its effect on BMI.

Methods: This was a health education intervention project. An initial session on the importance of adopting appropriate eating habits was conducted in April 2026. The intervention targeted children from 5 to 10 years old with overweight (BMI percentile ≥ 85 and < 95) or obesity (BMI percentile ≥ 95), and their respective caregivers. Approximately two months later, a follow-up session was conducted. BMI was assessed in the children at both time points. The normality of the differences was assessed using the Shapiro-Wilk test ($p = 0.65$), followed by a paired t-test. Parental feeding practices were assessed using the *Child Feeding Questionnaire* (CFQ).

Results: Of the 21 children initially recruited, 6 completed the follow-up (adherence rate: 28.6%). A reduction in mean BMI from 19.42 to 18.89 kg/m² and in BMI percentile from 91.67 (SD $\pm$ 4) to 80.33 (SD $\pm$ 14) was observed, corresponding to a mean change of $\Delta = -11.3$ percentage points, which did not reach statistical significance ($p = 0.055$). Furthermore, 4 of the 6 children moved into the healthy weight category following the intervention. At baseline, the CFQ indicated that parents were the main agents in child feeding, generally perceived their child's weight as normal, and reported moderate awareness and monitoring of unhealthy food intake, with some variability in concerns about current and future weight.

Conclusions: Although the results obtained did not reach statistical significance, an improvement in BMI was observed in several children, with some moving into the "healthy weight" category. These findings suggest that educational interventions on healthy eating may contribute to the adoption of healthier behaviours. Implementing such interventions continuously and on a larger scale may be relevant, but further studies involving larger samples and longer follow-up periods are needed.

MGF02

PRIMARY HYPERPARATHYROIDISM WITH RENAL INVOLVEMENT IN PRIMARY CARE - A CASE REPORT

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Background/Aims: Primary hyperparathyroidism (PHPT) is often asymptomatic, frequently detected incidentally through routine biochemistry (1). In primary care, a timely diagnosis is essential to prevent chronic end-organ complications such as nephrolithiasis and chronic kidney disease (CKD) (1,2). This case highlights when family doctors should suspect PHPT and how to initiate early assessment and management.

Methods: A 57-year-old female with controlled hypertension presented in November 2025 with an incidentally elevated serum creatinine of 1.5 mg/dL on pre-operative workup. She was advised to repeat the renal study in three months. In April 2026, repeat testing showed persistent renal impairment (creatinine 1.59 mg/dL; eGFR 37.7 mL/min/1.73m²), accompanied by elevated uric acid (9.1 mg/dL) and urea (81 mg/dL). Consequently, avoidance of nephrotoxic drugs (NSAIDs) was advised, and a systematic diagnostic workup was initiated to rule out secondary causes of kidney disease.

Results: The results revealed an ionized calcium of 6.7 mg/dL (reference: 4.44–5.20 mg/dL), and a severely elevated parathyroid hormone (PTH) of 1201 pg/mL (reference: 19–88 pg/mL). Renal ultrasound showed bilateral parenchymal thinning and multiple non-obstructing calculi, the largest measuring 11 mm on the left and 10 mm on the right. Thyroid ultrasound identified a 2.1 cm hypoechoic nodule posterior to the right thyroid lobe, highly suggestive of a parathyroid adenoma. Despite the absence of classic systemic hypercalcemic symptoms, the patient was referred to general surgery consult for parathyroidectomy, due to end-organ damage (bilateral nephrolithiasis and CKD stage 3b).

Conclusion: This case illustrates that asymptomatic PHPT can mask severe, silent end-organ damage. Family doctors must maintain a high index of suspicion, measuring calcium in patients with unexplained renal impairment, osteoporosis, or nephrolithiasis. Once PHPT is suspected, a prompt diagnostic cascade including PTH measurement, renal ultrasound, and kidney function assessment is crucial to identify surgical candidates and prevent irreversible renal damage (2).

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MGF03

STONES, BONES AND A HIDDEN GENE: SEVERE PRIMARY HYPERPARATHYROIDISM IN A YOUNG WOMAN - A CASE REPORT

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Objectives: Primary hyperparathyroidism (PHP) is typically a disease of postmenopausal women, but genetic forms are disproportionately represented among young patients, with up to 71% of hyperparathyroidism-jaw tumour syndrome (HPT-JT) cases recognised before the age of 30. We describe the case of a young woman with severe PHP, nephrolithiasis and unexpectedly low bone mineral density, in whom a pathogenic CDC73 variant was incidentally identified, to highlight the role of the family physician in early recognition, referral and long-term multidisciplinary follow-up.

Methods: Retrospective case report based on clinical records from general practice, urology, endocrinology, surgery and genetics consultations, covering a follow-up period of more than five years.

Results: A 23-year-old woman was admitted for an obstructive left ureteral calculus requiring stenting and stone extraction. Prompted by this episode of nephrolithiasis, her family physician requested serum calcium and parathyroid hormone (PTH) evaluation one year later, revealing marked hypercalcemia (total calcium 13.9-14.1 mg/dL) and elevated PTH (up to 493 pg/mL), consistent with PHP. Bone densitometry showed osteoporosis unexpectedly severe for her age. Cervical ultrasound and parathyroid scintigraphy localized a hyperfunctioning nodule inferior to the left thyroid lobe. She underwent left inferior parathyroidectomy; histology confirmed a parathyroid adenoma with associated thymic tissue. The postoperative course was complicated by transient hypocalcemia, left vocal cord palsy, which resolved within months with speech therapy, and Horner syndrome, which improved gradually over two years. Genetic testing was negative for MEN1 but identified an incidental heterozygous nonsense variant (c.358C>T) in CDC73, associated with HPT-JT and familial isolated hyperparathyroidism. Long-term follow-up shows adequate biochemical control but persistently reduced bone mineral density, and the patient's wish for future pregnancy raises further counselling needs regarding autosomal dominant transmission and screening of relatives.

Conclusions: Severe, early-onset PHP warrants genetic screening even without a suggestive family history, since identifying pathogenic variants such as CDC73 has direct implications for long-term surveillance, family counselling and reproductive planning. Family physicians play a pivotal role in triggering the diagnostic work-up from apparently unrelated complaints, such as recurrent nephrolithiasis, and in coordinating multidisciplinary long-term care.

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2. Gheorghe AM, Sima OC, Florescu AF, et al. Insights into Hyperparathyroidism-Jaw Tumour Syndrome: From Endocrine Acumen to the Spectrum of CDC73 Gene and Parafibromin-Deficient Tumours. Int J Mol Sci. 2024;25(4):2301.

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MGF04

SEVERE AUTOIMMUNE HYPOTHYROIDISM IN A YOUNG ADULT: AN UNEXPECTED DIAGNOSIS IN THE CONTEXT OF PALPITATIONS

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Introduction: Primary hypothyroidism is one of the most common endocrine disorders, with autoimmune thyroiditis being one of the leading causes in regions with adequate iodine intake. Its clinical presentation is usually insidious and non-specific and, in severe forms, may include multisystem manifestations such as fatigue, cold intolerance, constipation, weight gain, bradycardia and oedema, potentially progressing to myxoedema coma in extreme cases. However, some patients may present with a marked discrepancy between biochemical severity and clinical manifestations, making the diagnosis more challenging.

Case report: A 22-year-old man, previously healthy, presented to primary care with recently developed intermittent palpitations, without other symptoms suggestive of thyroid dysfunction. Due to persistent symptoms, he was assessed in the Emergency Department, where physical examination was essentially unremarkable and he remained haemodynamically stable. Electrocardiography showed sinus rhythm, and cardiovascular assessment revealed no evidence of acute pathology. Laboratory tests showed a TSH level of 382.59 μ IU/mL and free T4 below 5.4 pmol/L, associated with elevated transaminases and lactate dehydrogenase, with mild renal impairment. Despite the marked biochemical severity, he remained asymptomatic at that time, with no palpitations, bradycardia, hypothermia, altered mental status or other signs suggestive of myxoedema coma. Levothyroxine 150 μ g/day was initiated. Subsequent investigation revealed thyroid ultrasonographic findings consistent with thyroiditis and positive antithyroid antibodies, establishing the diagnosis of autoimmune hypothyroidism consistent with Hashimoto's thyroiditis. After several months, he discontinued treatment due to forgetfulness and because he remained asymptomatic, presenting again in November 2025 with marked biochemical deterioration (TSH 312.49 μ IU/mL; free T4 5.1 pmol/L), again without significant clinical manifestations. Following resumption of levothyroxine and subsequent dose adjustment to 175 μ g/day, progressive improvement was observed, with TSH 5.31 μ IU/mL and free T4 14.7 pmol/L in September 2026

Discussion and conclusion: This case highlights that severe biochemical hypothyroidism may present with limited clinical manifestations, even in a young adult, and that the absence of symptoms does not imply adequate disease control. The deterioration following treatment discontinuation reinforces the importance of treatment adherence and laboratory monitoring. Continuity of care and collaboration between primary care and Endocrinology were essential for diagnosis, follow-up and treatment adjustment.

Keywords: hypothyroidism; Hashimoto's thyroiditis; palpitations; young adult.

MGF05

PERSISTENT HYPONATRAEMIA AND HYPOTENSION IN AN OLDER PATIENT: A DIAGNOSTIC CHALLENGE OF CENTRAL ADRENAL INSUFFICIENCY

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Introduction: Central adrenal insufficiency may present insidiously and with non-specific symptoms, particularly in older adults. Manifestations such as fatigue, anorexia, hypotension and hyponatraemia may initially be attributed to more common causes, such as dehydration, gastrointestinal illness or adverse effects of medication. Persistent hyponatraemia despite correction of hypovolaemia should prompt consideration of this condition in the differential diagnosis.

Case report: An 83-year-old man, previously independent, with a history of hypertension and depressive disorder, was receiving lercanidipine, olmesartan, venlafaxine and trazodone. Following an initial episode of dizziness with transient loss of consciousness, he was admitted to the Emergency Department with diarrhoea, fatigue and severe hypotension (70/45 mmHg). Laboratory tests showed hyponatraemia (131 mmol/L), acute kidney injury and probable hypovolaemia. The clinical presentation was initially interpreted as dehydration secondary to gastrointestinal illness, potentially exacerbated by antihypertensive therapy. Despite intravenous fluid replacement, hyponatraemia persisted and worsened, reaching 121 mmol/L despite apparent euvolaemia. Reduction of antidepressant therapy did not lead to resolution. Given the persistence of electrolyte abnormality, serum cortisol was measured and was below the limit of detection (<27.6 nmol/L), with a low ACTH level (1.69 pg/mL), consistent with central adrenal insufficiency. Hypothalamic-pituitary magnetic resonance imaging revealed no significant structural abnormalities. Following initiation of intravenous hydrocortisone, there was marked clinical improvement and progressive normalization of serum sodium, and the patient was discharged on oral replacement therapy.

Discussion and conclusion: This case highlights the overlap between the clinical manifestations of adrenal insufficiency and common conditions in older adults, which may delay diagnosis. The persistence of hyponatraemia despite adequate volume replacement, was crucial in prompting reconsideration of the initial diagnosis and assessment of the hypothalamic-pituitary-adrenal axis. The absence of hyperkalaemia did not exclude the diagnosis, given the usual preservation of aldosterone secretion in central adrenal insufficiency. Longitudinal integration of clinical information, medication review and collaboration between primary and secondary care were essential for diagnosis and follow-up.

Keywords: central adrenal insufficiency; hyponatraemia; hypotension; cortisol deficiency.

MGF06

ASSESSMENT OF FOOD INSECURITY IN A POPULATION OF ADULTS WITH TYPE 2 DIABETES

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Background & Aim: Food insecurity (FI) is a social determinant of health with a potential impact on the management of type 2 diabetes (T2DM), as it may affect dietary quality, treatment adherence and the risk of metabolic complications. In primary healthcare (PHC), identifying food insecurity is essential for an integrated, person-centred clinical approach geared towards health equity.

Objectives: To assess the relationship between the prevalence and severity of food insecurity and metabolic control in adults with T2DM receiving care in PHC.

Methods: A cross-sectional observational study conducted between October and December 2025, with a convenience sample of 139 participants. Data were collected via face-to-face interviews and through the SClínico® system. Food insecurity was assessed using a validated instrument employed in the National Food and Physical Activity Survey (IAN-AF) – the Household Food Security Survey Module. sociodemographic variables, glycaemic control, lipid profile according to cardiovascular risk, blood pressure and renal function were analysed. Descriptive and inferential statistics were applied, with a significance level of 5%.

Results: The mean age was 70.4 ± 9.9 years (53.2% male), with a predominance of low educational attainment. The overall prevalence of FI was 29.5% (mild 23%, moderate 5.8% and severe 0.7%). No significant differences were observed between individuals with and without FI in terms of age, gender, educational attainment, HbA1c, LDL cholesterol. It was found that individuals with FI were more likely to have hypertriglyceridaemia (43.9%, $p=0.013$) and impaired renal function (21.4%, $p=0.013$), with these results being statistically significant ($p<0.05$).

Conclusions: FI was found to have a high prevalence in this population, much higher than that observed in the IAN-AF study, and was associated with alterations in the lipid profile, greater central adiposity and renal impairment. Its systematic identification in primary care settings may enable earlier, individualised and integrated interventions, promoting a diabetes management approach that takes social determinants of health into account.

MGF07

PETRIFIED EAR - A DIAGNOSTIC PEARL

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Background: Unusual complaints may be dismissed in the middle of the hustle of a Family Medicine Day. A sensation of a petrified ear could easily be one of those. However, rather than dismissing such patient's concern, the search for answers and ruling out an organic cause should be the next step.

Description (Methods/Results): This is the case of a 47-year-old Caucasian man with diagnosis of Raynaud's syndrome, chronic gastritis, venous insufficiency, and Carpal tunnel syndrome. His medication is nifedipine 60 mg. He had a previous history of inhaled cannabinoid and tobacco use and no known allergies. During an appointment, he reported a sensation of "petrification" of the left ear that had been present for over a year, even though it was painless. Physical examination revealed a left retroauricular swelling with no signs of inflammation. An ultrasound showed an "ovoid-shaped lesion measuring 7.5 x 2 mm, suggestive of a superficial reactive lymph node corresponding to the palpable swelling." However, screening for adrenal insufficiency, through cortisol and ACTH laboratory testing, actually revealed hypocortisolism (cortisol 2.1 µg/dL; ACTH 20.2 pg/dL). He was referred to an Endocrinology consultation for re-evaluation, in which adrenal insufficiency was confirmed. He successfully initiated treatment with oral hydrocortisone and maintains his follow-up.

Conclusion: The peculiarities of many medical diagnoses can be dismissed, particularly when dealing with rare diseases. Adrenal insufficiency may be part of this group of diseases, due to its nonspecific symptoms and low prevalence in the general population. Given the complexity and vast range of medical issues in Family Medicine consultation, sometimes it is difficult to be familiar with every detail of each disease; nevertheless, the effort to investigate and rule out hypotheses, even uncommon ones, remains essential.

10º CURSO DE ENFERMAGEM EM ENDOCRINOLOGIA

POSTERS | ENFERMAGEM

ENF01

CARING IN COMPLEXITY: NURSING INTERVENTION IN THE PATIENT WITH DIABETES AND A PACEMAKER

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Background/Aims: Type 2 diabetes mellitus (T2DM) is a major cardiovascular risk factor, frequently associated with multiple comorbidities and greater complexity of disease management. In the older patient with cardiovascular disease and an implantable cardiac device, nursing is central to clinical surveillance, control of risk factors, treatment adherence, and empowerment for self-care. This study aimed to describe the nursing intervention in a patient with T2DM and complex cardiovascular disease who underwent pacemaker implantation, highlighting the nursing contribution to the integrated management of cardiovascular and metabolic risk, patient safety, and the promotion of self-care.

Methods: Descriptive single-case study of a 79-year-old man with T2DM, arterial hypertension, dyslipidaemia, former smoking, heart failure, atrial fibrillation, and multivalvular disease. He presented to the Emergency Department with an episode of syncope and dizziness, and clinical and cardiological assessment established an indication for a permanent cardiac device; a dual-chamber DDD pacemaker was implanted. A multidimensional, individualised nursing assessment considered age, comorbidities, cardiovascular risk factors, diabetes, and the presence of the device, and guided an intervention comprising clinical and cardiovascular surveillance, control of cardiovascular risk factors, assessment of diabetes management and treatment adherence, education on healthy nutrition, lifestyles, and appropriate physical activity, surveillance of the implantation site, teaching on device-related care and recognition of warning signs, reinforcement of device follow-up, promotion of autonomy and self-care, and continuity of care across levels of assistance.

Results: The nursing intervention made it possible to identify individual education and empowerment needs, reinforce the patient's knowledge of diabetes, cardiovascular disease, and the implanted device, and promote self-care strategies appropriate to the clinical condition. The individualised approach contributed to increased safety, autonomy, and active patient participation in the management of his own health and supported the early identification of needs through the nursing consultation as a privileged setting for integrating surveillance, education, prevention, and empowerment.

Conclusion: The complexity of the patient with diabetes and cardiovascular disease requires an integrated, individualised, and person-centred nursing intervention. Through surveillance, health education, management of risk factors, and empowerment for self-care, the nurse plays a decisive role in promoting safety, autonomy, and continuity of care. Caring in complexity means caring for the person beyond diagnosis, integrating disease, treatment, device, context, and the capacity for self-care.

Keywords: Type 2 diabetes mellitus; cardiovascular disease; Pacemaker; Nursing; Cardiovascular risk; Self-care; Health education.

ENF02

MAPPING THE MULTIDIMENSIONAL NEEDS OF PEOPLE WITH TYPE 2 DIABETES MELLITUS DURING THE TRANSITION TO RETIREMENT: A SCOPING REVIEW

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Objectives: The transition to retirement can modify daily routines, social roles, identity, health behaviours and access to healthcare, with potential consequences for diabetes self-management. This scoping review aimed to map and synthesise the multidimensional needs of adults and older adults with type 2 diabetes mellitus (T2DM) during the transition to retirement, and to identify barriers, facilitators and knowledge gaps.

Material and Methods: A scoping review was conducted according to Joanna Briggs Institute methodology and reported in accordance with PRISMA-ScR. MEDLINE (via PubMed), CINAHL, Scopus, Web of Science and RCAAP were searched, together with grey literature sources. Studies published in English, Portuguese or Spanish were eligible without date restrictions. Eligibility was defined using the Population–Concept–Context framework. Study selection and data extraction were undertaken independently by two reviewers, and findings were synthesised narratively.

Results: The search identified 1,365 records; after removal of 302 duplicates, 1,063 records were screened and 10 studies were included. Most studies were quantitative and addressed retirement indirectly through outcomes including physical activity, medication adherence, multimorbidity, diabetes-related complications, work disability, early exit from employment, quality of life and perceived health. Needs were mapped across six domains: physical and clinical, self-management, psychological, social, work-related and spiritual. Physical and clinical, self-management and work-related needs were most frequently represented, whereas psychological and social dimensions were less consistently explored. No included study explicitly examined spiritual needs. Key barriers included complications, multimorbidity, disability, disruption of routines, medication non-adherence, healthcare access difficulties and socioeconomic inequalities. Facilitators included continuity of care, healthy behaviours, supportive environments and opportunities to reorganise daily routines after retirement.

Conclusions: Evidence on T2DM during the transition to retirement remains limited and fragmented. Retirement may represent both a period of vulnerability and an opportunity for health promotion and reorganisation of self-management. Anticipatory, person-centred approaches should address clinical status, self-management, psychological adaptation, social support, work-related circumstances and continuity of care before, during and after retirement. The absence of evidence on spiritual needs and the scarcity of targeted nursing interventions identify priorities for future longitudinal, qualitative and intervention research.

ENF03

INTERNAL REFERRAL PROTOCOL FOR PEOPLE WITH DIABETES RECEIVING INJECTABLE THERAPY: A NURSING CARE PATHWAY

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Objectives: To develop and standardize an internal nursing referral pathway for people with diabetes receiving insulin or other injectable therapy, promoting safe administration, therapeutic education, clinical monitoring, early identification of complications, and continuity of care.

Material and Methods: A practice-based organizational protocol was developed to structure internal nursing referral and follow-up. The pathway includes initial assessment, individualized care planning, therapeutic education, monitoring, multidisciplinary coordination, clinical documentation, and continuous improvement. The initial assessment addresses clinical history, treatment adherence, and difficulties related to injectable therapy administration. Therapeutic education covers injection technique, medication storage, rotation of injection sites, warning signs of hypoglycemia and hyperglycemia, healthy lifestyles, and adherence to the dietary plan. Follow-up includes assessment of glycemic status, capillary glucose records, complementary examinations, treatment adherence, and early signs of complications. Nursing interventions are documented in the clinical record and coordinated with medical and other healthcare professionals whenever required.

Results: The resulting protocol established a seven-step internal care pathway: (1) identification and referral of eligible people; (2) comprehensive nursing assessment; (3) therapeutic education and supervised training; (4) individualized care planning; (5) regular monitoring and follow-up; (6) multidisciplinary coordination; and (7) reassessment and continuous improvement. The pathway also standardizes documentation of nursing interventions, updating of care plans, monitoring of diabetes-related indicators, periodic protocol review, and continuing education of the nursing team. It provides a common framework for identifying administration difficulties, reinforcing self-management skills, recognizing warning signs and complications, and supporting timely communication between professionals.

Conclusions: A structured internal nursing referral pathway can organize care for people with diabetes receiving injectable therapy by integrating assessment, education, technique supervision, monitoring, documentation, and multidisciplinary coordination. Standardizing these procedures may strengthen safety, self-management support, and continuity of care. Future evaluation should assess implementation through predefined process indicators and subsequently examine clinical and patient-reported outcomes

ENF04

DIABETES AND OOCYTE QUALITY: METABOLIC CHALLENGES AND THE ROLE OF NURSING IN MAR

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Introduction: Diabetes Mellitus (DM) is a chronic metabolic disease with an increasing impact on women of reproductive age. Hyperglycaemia, insulin resistance, and associated hormonal alterations may compromise ovarian function, oocyte quality, and the outcomes of Medically Assisted Reproduction (MAR) techniques.

Objective: To analyse the influence of diabetes on oocyte quality and MAR outcomes, identifying the underlying pathophysiological mechanisms and highlighting the role of nursing in metabolic risk management.

Methodology: A narrative review of the scientific literature on diabetes, metabolism, ovarian function, and MAR was conducted. This was complemented by the consultation of national documents and reports on diabetes and medically assisted reproduction.

Results: Evidence suggests that hyperglycaemia and insulin resistance may alter the follicular microenvironment, promote oxidative stress and mitochondrial dysfunction, and compromise oocyte maturation and quality. The scarcity of national data makes it difficult to characterise this issue in Portugal. At the national level, there is an “epidemiological gap” due to the lack of publicly available and centralised statistical data on women with diabetes undergoing MAR. “Patient motivation and active engagement” are key factors for treatment adherence, metabolic control, and successful completion of the different stages of MAR. In this context, nursing plays an essential role in health education, promoting motivation and self-care, metabolic monitoring, surveillance during ovarian stimulation, and emotional support.

Conclusion: Adequate metabolic control, active patient participation, and individualised nursing interventions may contribute to optimising reproductive outcomes. The creation of registries incorporating metabolic variables could help address the current epidemiological gap and support future research.

Keywords: Diabetes Mellitus; Oocyte Quality; Health Education

ENF05

SPECIALIZED PEDIATRIC NURSING IN CHILDHOOD OBESITY: INTERVENTION STRATEGIES AND CLINICAL OUTCOMES

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Introduction: Childhood obesity is a chronic, multifactorial, and progressive disease. Complications emerge early, affecting metabolic, cardiovascular, and psychosocial dimensions. Structured, early, and continuous interventions are crucial.

Objectives: To evaluate the impact of a specialized nursing clinic over 24 months on the anthropometric and behavioral parameters of children and adolescents (aged 8–18).

Methods: Longitudinal observational study involving 3 non-pharmacological nursing consultations. Initial assessment: Anthropometrics (BMI, body fat), lifestyle, and eating habits. Comorbidities screened: Dyslipidemia, hypertension, steatosis, and glycemic alterations. Core intervention: Individualized education, SMART goals, and family involvement. Key pillars: Meal regularity, ultra-processed food reduction, gradual physical activity, screen time reduction, and emotional coping strategies.

Results: Behavioral: Significant improvement in dietary habits, increased physical activity, and reduced sedentary screen time.

Anthropometric: Stabilization or reduction of BMI alongside favorable body composition trends. Sustainability: Progress was maintained even in the presence of baseline metabolic comorbidities. Continuity: Consistent follow-up and positive reinforcement proved to be the primary drivers of behavioral change.

Conclusions: Specialized nursing consultations are essential for the effective management of pediatric obesity. Evidence-based, structured care successfully empowers families and fosters sustainable health habits.

Key message: Specialized nurses look beyond body weight, guiding individuals and families toward healthier life trajectories.

ENF06

ARTIFICIAL INTELLIGENCE AND SELF-MANAGEMENT IN TYPE 2 DIABETES: SIMULATED CLINICAL CASE OF NURSING INTERVENTION IN THE ERA OF CHATGPT

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Introduction: People with type 2 diabetes mellitus (T2DM) increasingly arrive at consultations with information generated by artificial intelligence (AI). Evidence shows modest benefits of AI tools and continuous glucose monitoring (CGM) in self-management, but also safety limitations and an effect that fades without structured follow-up.

A simulated clinical case, built on evidence for educational purposes, illustrates the nurse's role in safely integrating these tools.

Objectives: Illustrate a simulated nursing intervention model that integrates generative AI into the self-management of T2DM, demonstrating how structured therapeutic education and supervised AI use can improve clinical, behavioral, and digital literacy outcomes.

Methodology: This is a simulated case study, developed for pedagogical purposes and grounded in current evidence on T2DM self-management and the safe integration of AI tools into nursing practice. Simulated Clinical Case Male, 64 years old, T2DM with 13 years of progression, HbA1c 9.2%, BMI 31.4 kg/m², low digital literacy (eHEALS 14/40), followed in an Endocrinology clinic. He used unsupervised generative AI, obtaining content inappropriate for his risk profile.

Results: The 12-week intervention led to measurable improvements across clinical, behavioral, and digital literacy domains. HbA1c decreased from 9.7% to 8.3%, and time in range increased from 38% to 61%. Self-management and knowledge scores improved to ≥ 4 , indicating strengthened behavioral competencies. Digital literacy also increased substantially, with eHEALS rising from 14 to 26/40, marking a transition from low to moderate literacy. The patient shifted from unsupervised to supervised use of generative AI, demonstrating safer and more effective integration of digital tools into diabetes self-management.

Conclusions/Implications: AI and technology are already part of patients' self-management, with or without professional supervision. This case illustrates a replicable model of nursing curati on, which evidence suggests is crucial to transform technological stimulus into sustained self-m anagement.

Keywords: type 2 diabetes mellitus; artificial intelligence; self-management; therapeutic education.

ENF07

THERAPEUTIC SUMMER CAMPS: IMPACT ON AUTONOMY AND METABOLIC CONTROL IN TYPE 1 DIABETES MELLITUS

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Background: Adolescence is a critical period for the management of Type 1 Diabetes Mellitus (T1DM), requiring innovative educational approaches that promote autonomy and adherence to treatment. Therapeutic summer camps provide a non-formal environment for health education, where the continuous presence of Specialized Pediatric Nurses ensures clinical safety and supports self-management development.

Aim: To evaluate the impact of a therapeutic summer camp on metabolic control and self-efficacy among adolescents with T1DM, highlighting the contribution of specialized nursing interventions.

Methods: A descriptive longitudinal study was conducted involving 21 adolescents with T1DM aged 12–17 years (52.4% female; mean age 14.1 ± 1.6 years). Glycated hemoglobin (HbA1c) was assessed before the camp and during two follow-up consultations. Participants completed a semi-structured questionnaire exploring satisfaction, perceived benefits, challenges, and the role of healthcare professionals.

Results: Metabolic outcomes demonstrated a progressive improvement, with mean HbA1c decreasing from 7.42% before the intervention to 7.00% at the second follow-up consultation. The greatest individual improvement was observed in a participant whose HbA1c decreased from 11.1% to 7.8%. All participants (100%) reported that the camp met their expectations and emphasized the benefits of peer interaction, normalization of their condition, and the sense of safety provided by continuous nursing support. Initial social integration and the management of intense physical activity were identified as the main challenges. Almost half of the participants (45%) suggested extending the duration of the camp.

Conclusions: Therapeutic summer camps supervised by Specialized Pediatric Nurses are valuable interventions for adolescents with T1DM, contributing to improved metabolic control, enhanced self-management skills, and greater emotional well-being. These findings reinforce the importance of integrating experiential and peer-supported educational strategies into diabetes care programs.

14TH ADVANCED COURSE OF ENDOCRINOLOGY

ORAL COMMUNICATIONS AND CLINICAL CASES

CO01

PRIMARY HYPERPARATHYROIDISM IN PREGNANCY WITH PERSISTENT DISEASE AFTER SURGICAL TREATMENT

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Introduction: Primary hyperparathyroidism during pregnancy is rare. Most cases are mild and can be successfully managed conservatively. The risk of adverse pregnancy outcomes appears to correlate with maternal serum calcium levels. In more severe cases, management is challenging due to the lack of approved pharmacological therapies and limited evidence supporting the safe off-label use of available agents. Therefore, surgery represents the most feasible treatment option and should ideally be performed during the second trimester.

Case presentation: A 38-year-old woman was diagnosed with normocalcemic primary hyperparathyroidism without evidence of end-organ involvement. Neck ultrasound identified a 15-mm hypoechoic lesion posterior and inferior to the right thyroid lobe, with concordant mild uptake on parathyroid scintigraphy. Before surgery could be considered, the patient had an unplanned pregnancy. At 8 weeks and 6 days of gestation, asymptomatic hypercalcemia was documented for the first time, with corrected calcium of 10.8 mg/dL (<10.2 mg/dL). Conservative management with oral hydration, a low-calcium diet, and furosemide was initiated. Despite this, calcium increased to 11.2 mg/dL at 11 weeks and 3 days, prompting intravenous fluids and furosemide. A transient improvement was observed (10.4 mg/dL), followed by a subsequent rise to 11.2 mg/dL at 15 weeks and 3 days. Given persistent hypercalcemia (1 mg/dL above normal), ultrasound-guided parathyroidectomy was performed at 18 weeks and 4 days of gestation. Intraoperative PTH decreased by 58.4% and 78.3% at 5 and 15 minutes, respectively. The procedure was uneventful, and calcium normalized by postoperative day 14 (20 weeks and 4 days of gestation). Histopathology revealed a 7-mm parathyroid lesion without a clear capsule, favoring hyperplasia. Genetic testing showed no pathogenic germline variants in MEN1 or RET. Calcium levels remained normal for most of the pregnancy, with a mild increase at 37 weeks (10.4 mg/dL). Delivery occurred at 40 weeks and 6 days with vacuum assistance, without immediate maternal or neonatal complications. During lactation, hypercalcemia recurred five months postpartum (calcium 10.7 mg/dL).

Conclusion: Management of primary hyperparathyroidism during pregnancy can be challenging and requires a multidisciplinary approach. In cases of persistent hypercalcemia despite conservative treatment, second-trimester parathyroidectomy should be considered to improve maternal and fetal outcomes.

CO02

PREGNANCY IN A PATIENT WITH METASTATIC APPENDICEAL NEUROENDOCRINE TUMOUR: A RARE AND CHALLENGING CLINICAL CASE

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BACKGROUND: Appendiceal neuroendocrine tumours (appnets) are rare neoplasms, usually diagnosed incidentally following appendicectomy for acute appendicitis. Metastatic disease from appnets is exceptionally rare, and a coexisting midgut net should be considered. Pregnancy in women with metastatic nets is uncommon and presents clinical challenges due to limited evidence to guide management.

CASE REPORT: A 35-year-old woman was diagnosed at the age of 23 years with a non-functioning grade 2 appendiceal net (2 mitoses/2 mm²; ki-67 index 3–20%) and synchronous metastases (pt2nmx1) following appendicectomy for acute appendicitis in 2014. At our institution she underwent extensive cytoreductive surgery, including right hemicolectomy, omentectomy, left salpingo-oophorectomy and multiple peritonectomies, with histopathological confirmation of metastatic g2 appnet. Post-operative imaging demonstrated small residual hepatic/perihepatic lesions with somatostatin receptor (sstr) expression, and somatostatin analogues (ssas) were initiated. Serial imaging showed no significant disease progression. In 2019, the patient conceived spontaneously and opted to discontinue ssas at 5 weeks' gestation. Pregnancy was uneventful, and a healthy male infant was delivered at term by caesarean section for arrest of labour. Ssas were resumed after delivery. Post-partum imaging confirmed persistent low-volume disease without new metastatic lesions. Subsequent ct,sstr-pet and liver mri demonstrated radiological stability. Prrt was considered, however, its role in this setting is not yet established as standard of care. Moreover, the disease has remained radiologically stable to date.

Conclusion: Metastatic appnets are exceptionally rare, and pregnancy requires an individualised multidisciplinary approach. This case highlights the potential role of extensive cytoreductive surgery in achieving durable disease control and illustrates that temporary ssa withdrawal during pregnancy may be feasible in selected patients with non-functioning disease, without clinically significant disease progression. Although current evidence suggests a favorable maternal and fetal safety profile for ssas during pregnancy, data on their withdrawal remain limited, further reports are needed to guide treatment decisions in women with metastatic nets.

CO03

ACROMEGALY IN PREGNANCY: LONG-TERM CLINICAL OUTCOMES OF A 27-YEAR FOLLOW-UP CASE

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Background/aims: Acromegaly during pregnancy is a rare clinical scenario requiring careful management regarding tumor growth, therapeutic safety, and hormone monitoring. This case highlights the 27-year longitudinal management of a female patient, emphasizing maternal-fetal safety and long-term comorbidity surveillance.

Case report: a 22-year-old female presented in 1999 with acral overgrowth, facial coarsening, hyperhidrosis and headaches. Initial evaluation revealed a 2-cm pituitary macroadenoma with igf-1 982 ng/ml (over 3 times the upper limit of normal) and gh 142 ng/ml. She underwent transsphenoidal surgery, with histopathology confirming a gh- and prolactin-immunoreactive adenoma. Due to persistent postoperative hypersecretion, combination medical therapy with somatostatin receptor ligands (srls) and dopamine agonists was initiated, achieving biochemical control. She achieved two spontaneous pregnancies (2008 and 2013), with medical therapy being withheld upon diagnosis. Serial igf-1 levels remained low to normal, consistent with estrogen-mediated hepatic gh resistance. Visual fields and maternal status remained stable without tumor expansion. She completed two full-term pregnancies via cesarean section, delivering healthy neonates. Postpartum, srl therapy was resumed. Over 27 years of follow-up (1999–2026), magnetic resonance imaging showed evolution to secondary empty sella with a stable 9.5-mm residual microadenoma. Systematic screening confirmed the absence of colonic polyps, normal sleep study, and stable minor valvular regurgitation with preserved ejection fraction.

Conclusion: this case illustrates that favorable maternal-fetal outcomes can be achieved in acromegaly with temporary srl withdrawal. The observed hormonal stability during gestation likely reflects estrogen-induced hepatic resistance to gh action. Long-term combination medical therapy successfully controlled tumor burden, while structured comorbidity surveillance ensured overall maternal health across nearly three decades.

CO04

BEYOND TYPE 1 DIABETES: MULTIDISCIPLINARY EVALUATION AS THE KEY TO A DIAGNOSIS OF WOLFRAM SYNDROME UNMASKED DURING PREGNANCY

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Background: Wolfram syndrome (WS) is a rare autosomal recessive neurodegenerative disorder. Its most frequent form, Wolfram syndrome 1 (classical), is caused by biallelic WFS1 variants and characterized by diabetes mellitus, optic atrophy, deafness, and diabetes insipidus. The diabetes is insulin-deficient and autoantibody-negative and is frequently misclassified as type 1 diabetes mellitus (T1DM) as it is usually the first manifestation. Optic atrophy is the most useful discriminating feature. Pregnancy in WS is rarely reported and poses distinct risks.

Case Presentation: A 25-year-old woman diagnosed with T1DM at age 5 and treated with insulin since, with poor long-term glycemic control due to non-adherence, presented at 17 weeks and 2 days of a dichorionic diamniotic pregnancy to an Endocrinology–Obstetrics consultation. Multidisciplinary review prompted ophthalmologic referral, which revealed bilateral optic disc pallor (optic atrophy) not attributable to diabetic retinopathy. C-peptide was low, but islet autoantibodies had never been tested, so autoimmune T1DM had never been confirmed. Targeted genetic testing identified two WFS1 variants compatible with WS—one of uncertain significance and one likely pathogenic—though results became available only after delivery. The phenotype of insulin-deficient diabetes without documented autoimmunity, combined with optic atrophy, strongly supports a causative role. During pregnancy, glycemic control remained suboptimal owing to low adherence, but HbA1c improved from 9.3% at first visit to 7.3% at 32 weeks and 2 days gestation. Delivery was by elective cesarean section at 33 weeks and 2 days due to abnormal fetal Doppler in one twin. Two newborns were delivered—male 1950 g (35th percentile) and female 1820 g (28th percentile)—without peripartum or postpartum complications. Since delivery, the patient reports no symptoms of diabetes insipidus, no neurologic or urologic complaints and awaits otorhinolaryngology and neurology assessment to exclude other disease-related manifestations.

Discussion: This case illustrates how routine multidisciplinary follow-up with ophthalmologic evaluation reframed presumed T1DM toward a clinical diagnosis of WS, supported by a compatible WFS1 variant of not-yet-proven pathogenicity, altering prognosis, surveillance, and genetic counseling. Insulin-deficient diabetes without documented autoimmunity should prompt consideration of monogenic and syndromic causes, with extrapancreatic signs such as optic atrophy playing a pivotal role in guiding diagnosis.

CO05

HYPERCALCEMIA ASSOCIATED WITH CYP24A1 MUTATION IN PREGNANCY: A CASE REPORT

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Introduction: cyp24a1 gene encodes 24-hydroxylase, an enzyme involved in vitamin d catabolism. homozygote mutations of cyp24a1 lead to 1,25(oh)vitamin d excess and subsequent hypercalcemia, with low serum parathyroid hormone. pregnancy further increases active vitamin d, making affected women particularly susceptible to hypercalcemia, which poses significant risks to both mother and fetus.

Case Report: a 32-year-old woman, 12 weeks pregnant, was referred to endocrinology for gestational diabetes (fasting glycemia: 94mg/dl). she had a history of idiopathic infantile hypercalcemia, cyp24a1 mutation, nephrocalcinosis and familial history of the same mutation in both parents. before pregnancy her calcium levels were normal. at 10 weeks' gestation her serum calcium was 10.8mg/dl [reference range (rr): 8.8-10.6] and at 16 weeks it rose to 11.3mg/dl. labs showed normal albumin levels, low magnesium (1.4mg/dl, rr: 1.9-2.5), low-normal phosphate (2.2-2.8mg/dl, rr: 2.5-4.5), normal 25(oh)vitamin d (35ng/ml, rr: 30-100), high 1,25(oh)vitamin d3 (> 150pg/ml, rr: 18-90) and undetectable parathyroid hormone levels; calciuria was elevated (255mg/24h, rr: <250). she was advised to avoid sun exposure, limit calcium-rich foods, drink >2l of water daily, and begin magnesium supplementation. by 20 weeks, her calcium levels reached 12.1mg/dl, necessitating hospitalization for intravenous fluids, which lowered her calcium to 10.7 mg/dl. however, calcium levels rebounded to 12.6mg/dl two weeks later, requiring a second short hospitalization for fluid therapy. her calcium gradually declined, reaching normocalcemia (10.6mg/dl) by 27 weeks, which was maintained throughout the third trimester. magnesium supplementation was increased up to 8 sachets daily due to persistent hypomagnesemia. gestational diabetes remained well-controlled with lifestyle measures only. fetal ultrasounds were normal. delivery occurred without complications at 39 weeks, with a healthy newborn and normal calcium levels in both.

Conclusions: cy24a1 mutation is a rare cause of hypercalcemia in pregnancy, with fewer than 30 cases reported globally. our patient experienced moderate hypercalcemia, requiring intravenous fluid therapy during the second trimester, the most critical period in pregnancies complicated by this mutation. management was challenging due to the limited therapeutic options in pregnancy. careful monitoring was essential for optimizing maternal and fetal outcomes.

CO06

MATERNAL-FETAL OUTCOMES IN PREGNANT WOMEN WITH TYPE 1 DIABETES ON HYBRID CLOSED-LOOP SYSTEMS: MEDTRONIC MINIMED 780G VERSUS TANDEM T:SLIM X2 CONTROL-IQ

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Background: Hybrid closed-loop (HCL) systems are increasingly used in pregnancy complicated by type 1 diabetes mellitus (T1DM), yet comparative data across platforms during gestation remain scarce. We aimed to compare glycemic control and maternal-fetal outcomes between pregnant women with T1DM treated with the Medtronic MiniMed 780G (MM780G) and the Tandem t:slim X2 Control-IQ systems (CIQ).

Methods: Single-center, retrospective observational study of T1DM pregnant women, followed at our centre. HbA1c and continuous glucose monitoring parameters were compared by trimester, together with obstetric and neonatal outcomes. Mann-Whitney and Fisher exact tests were applied. Given the small sample size, findings are exploratory and hypothesis-generating rather than confirmatory.

Results: Seventeen pregnant women with T1DM (5 on the MM780G and 12 on the CIQ) were analyzed. Maternal age (34 years [29-37]), pre-pregnancy BMI (23.5 kg/m² [22.5-26.4]), T1DM duration (21 years [11-30]) and pre-pregnancy HbA1c (MM780G 6.8% [6.5-7.3] vs CIQ 6.4% [6.0-7.0]; p=0.24). were comparable between groups. Glycemic control did not differ significantly between devices in any trimester (third-trimester pregnancy-specific time in range (TIRp, 63-140 mg/dL): 55% [52-56] vs 57% [48-61], p=0.47; HbA1c 6.0% [6.0-6.5] vs 5.8% [5.7-6.4], p=0.35, in MM780G and CIQ respectively). Cesarean delivery was frequent (5/5 vs 10/12). Neonates in the 780G group had higher birthweight (4010 g [3700-4020] vs 3335 g [2970-3568]; p=0.018), with a non-significant trend toward higher birthweight percentile (92 [85-96] vs 73 [55-86]; p=0.082) and higher LGA rates (60% vs 25%; p=0.28). Preterm birth occurred in 4/12 pregnancies in the CIQ group and in none of the MM780G group (p=0.26), two cases attributed to pre-eclampsia.

Conclusions: Glycemic control was comparable between the two HCL systems throughout pregnancy, although TIRp remained below the recommended target in both. Higher absolute birthweight in the MM780G group was partly explained by higher preterm birth rate in the CIQ group. Higher LGA rate in the MM780G group was not attributable to glycemic control, which was similar between groups throughout pregnancy, it may instead reflect factors such as maternal gestational weight gain, as reported in other HCL cohorts. These findings are hypothesis-generating and warrant confirmation in larger, multicenter studies.

CO07

GCK-MODY IN PREGNANCY: A CASE SERIES

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Background: Glucokinase maturity-onset diabetes of the young (GCK-MODY) is one of the most common forms of monogenic diabetes, and generally requires no treatment outside pregnancy. In pregnancy, management depends on fetal genotype. Identifying GCK-MODY before or early in pregnancy, and determining timing of insulin initiation, remain challenging. We aimed to describe the clinical characteristics, timing of diagnosis, hyperglycemia management, and perinatal outcomes of 5 pregnant women with GCK-MODY.

Methods: Retrospective case series of 5 pregnant women with confirmed GCK-MODY. We collected pre-conception, trimester-specific and postpartum HbA1c, as well as antidiabetic therapy, timing of GCK-MODY diagnosis, suspicion of fetal macrosomia, birthweight, and fetal genotype when available. Given the small sample, data are presented descriptively, case by case.

Results: Two patients had a pre-gestational diagnosis. In the remaining three, GCK-MODY was suspected during pregnancy and confirmed by genetic testing—at 36 weeks of gestation in one and postpartum in two. All patients had good glycemic control before pregnancy (HbA1c <6.5%). All started basal insulin during pregnancy (two in the first trimester, three in the second), and all but one progressed to a basal-bolus regimen. Glycemic control was satisfactory, improving in early pregnancy and remaining relatively stable in the second and third trimesters. Chorionic villus sampling (CVS), performed in one case at 14 weeks 4 days, showed that the fetus had not inherited the maternal GCK variant; this fetus and one other had suspected macrosomia. The former was a large-for-gestational-age (LGA) male neonate (3870 g, 91st percentile) and the latter a female neonate with normal birthweight (3660 g, 76th percentile). The other three women had no suspected macrosomia: two delivered normal-birthweight neonates (3105 g and 2770 g; 23rd and 16th percentiles) and one an LGA female neonate (3505 g, 92nd percentile). All neonates were full-term, with uncomplicated neonatal courses.

Table 1. Individual characteristics of 5 pregnancies

	Age	BMI	Timing of diagnosis (relative to pregnancy)	HbA1c before pregnancy	HbA1c 1 st T	HbA1c 2 nd T	HbA1c 3 rd T	Gestational age of insulin initiation	Suspected fetal macrosomia	Gestational age of delivery	Birthweight (g, percentile)	Fetal genotype
1	35	23.7	Before	6.4	6.5	5.5	6.4	10w+6d	No	39w+3d	3105, 23	Declined CVS
2	39	21.1	After	-	-	5.6	5.8	16w+0d	No	38w+6d	2770, 16	NA
3	34	30.7	During	5.7	5.9	5.4	5.9	10w+6d	Yes	39w+3d	3660, 76	NA
4	27	18.9	Before	6.3	6.2	5.6	6.2	17w+3d	Yes	38w+4d	3870, 91	Non-carrier
5	28	27.0	After	-	6.4	6.0	6.4	18w+5d	No	37w+3d	3505, 92	NA

Conclusion: Most women lacked a pre-conceptional diagnosis, illustrating the difficulty of recognizing this entity. Management varied between patients, reflecting the challenge of whether and when to start insulin therapy. These cases underline the importance of considering GCK-MODY in the differential diagnosis of hyperglycemia in pregnancy, as it profoundly affects glycemic management and fetal development

14TH ADVANCED COURSE OF ENDOCRINOLOGY

E-POSTERS

PO01

FETAL GENOTYPE-GUIDED INSULIN THERAPY IN A PREGNANCY COMPLICATED BY GLUCOKINASE-MODY (MODY 2): A CASE REPORT

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Background: Glucokinase maturity-onset diabetes of the young (GCK-MODY) is among the most common forms of monogenic diabetes, causing mild, non-progressive fasting hyperglycemia that generally requires no treatment outside pregnancy. In pregnancy, management depends on whether the fetus has inherited the maternal GCK variant. Invasive fetal genotyping by chorionic villus sampling (CVS) provides certainty but carries procedure-related risk. Serial ultrasound remains an unvalidated surrogate and non-invasive prenatal testing of the fetal GCK genotype from maternal cell-free DNA has demonstrated diagnostic accuracy and feasibility.

Case Presentation: A 27-year-old woman with a molecularly confirmed pathogenic GCK variant, diagnosed at age 16 on the basis of mild, stable fasting hyperglycemia (fasting glucose 133 mg/dL; HbA1c 6.5%), presented for pre-conception counseling. At that visit she was taking metformin 850 mg once daily with an HbA1c of 6.2% and was deemed fit for pregnancy. Nine months later, she presented to a combined Endocrinology–Obstetrics consultation at 12 weeks 3 days gestation (HbA1c 6.2%). CVS was performed at 14 weeks 4 days and showed the fetus had not inherited the maternal GCK variant, identifying a fetus at risk of accelerated growth from maternal hyperglycemia. Basal insulin was initiated at 17 weeks 3 days, with subsequent transition to a basal-bolus regimen at 20 weeks 3 days. Fetal growth was monitored by serial ultrasound. In the third trimester, macrosomia was suspected (estimated fetal weight 1964 g, 91st percentile) despite acceptable glycemic control (HbA1c 6.2%, 5.6%, and 6.0% in the first, second, and third trimesters, respectively). Spontaneous vaginal delivery complicated by shoulder dystocia occurred at 38 weeks 4 days, yielding a large-for-gestational-age (LGA) male neonate (birthweight 3870 g, 91st percentile). The neonatal course was uncomplicated. Postpartum, the patient required no antidiabetic treatment (HbA1c 5.9% at 56 days).

Discussion: In GCK-MODY pregnancy the fetal genotype is the rational basis for the treatment decision, and a definitive genotype-negative result supports maternal insulin therapy to mitigate the risk of macrosomia and LGA. However, macrosomia may still occur despite appropriate therapy and acceptable glycemic control. This case reinforces the value of preconception diagnosis, genetic counseling and structured newborn assessment.

PO02

PREGNANCY INDUCED/GESTATIONAL ARGININE VASOPRESSIN-RELATED DISORDER ASSOCIATED WITH HELLP SYNDROME

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Introduction: Pregnancy-induced/gestational arginine vasopressin-related disorder (gAVP-D) is a rare complication of pregnancy, presenting with hypotonic polyuria and polydipsia. It is related to excessive vasopressinase activity, an enzyme metabolised in the liver, that degrades arginine vasopressin, leading to impaired renal water reabsorption.

Case report: A 31-year-old woman at 37+2 weeks gestation (G2P1), presented to the emergency department with lower back pain, nausea, vomiting, fever and epistaxis. She also reported polydipsia, with a water intake of approximately 9 liters/day associated with polyuria for two weeks. Her medical history was notable for migraine and the fetus had previously been diagnosed with megacystis, which resolved spontaneously. The patient was hemodynamically stable and afebrile. Laboratory evaluation revealed leukocytosis and thrombocytopenia. Peripheral blood smear demonstrated target erythrocytes. Serum glucose was 68 mg/dL, creatinine 1.84 mg/dL(0.51-0.95), sodium 141 mmol/L(136-145). Liver function tests showed: total bilirubin 6.14 mg/dL(0.10-1.10), aspartate aminotransferase 132 U/L(4-27), alanine aminotransferase 203 U/L(4-34) and alkaline phosphatase 519 U/L(35-104). Coagulation studies revealed an increased INR. Cesarean was performed and a male newborn was delivered, weighing 2760 g, with Apgar scores of 7/8/9 at 1, 5, and 10 minutes. The obstetric team considered HELLP syndrome the most probable diagnosis. Endocrinology was consulted due to persistent polyuria and polydipsia. Although the patient reported clinical improvement, she continued to consume 3–4 L/day of fluids and had increased urine output of 4-6 L/day. Laboratory evaluation showed: sodium 138 mmol/L, blood osmolality 281 mOsm/KgH₂O(275-295), urine specific gravity: 1.004(1.005-1.030) and urine osmolality 195 mOsm/KgH₂O. Given unrestricted water access, clinical stability and biochemical findings, desmopressin therapy was not initiated. The patient remained under follow-up, with progressive improvement. She was discharged and, 18 days postpartum, reported normal fluid intake with complete resolution of symptoms. These findings supported the diagnosis of gAVP-D associated with HELLP syndrome. Further investigation was not considered necessary and no long-term endocrine follow-up was required.

Conclusions: This case highlights the importance of recognizing polyuria and polydipsia in pregnancy, which should prompt evaluation, including liver function assessment, allowing timely diagnosis and management of gAVP-D to reduce maternal and fetal morbidity.

POO3

SEVERE HYPERTRIGLYCERIDEMIA IN PREGNANCY: A CASE REPORT

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Introduction: Although triglyceride levels physiologically increase during pregnancy, severe gestational hypertriglyceridemia (>1000 mg/dL) is a rare and life-threatening condition associated with acute pancreatitis and adverse maternal-fetal outcomes, such as gestational diabetes mellitus, preeclampsia, preterm delivery or macrosomia. Managing hypertriglyceridemia during pregnancy can be challenging because the use of standard lipid-lowering pharmacotherapies is limited due to fetal safety concerns. This case report outlines the diagnosis and clinical management of severe hypertriglyceridemia in a pregnant patient in the third trimester.

Case presentation: A 39-year-old woman at 31 weeks of gestation (conceived via intracytoplasmic sperm injection) was noted to have lipemic serum on routine blood work, with a triglyceride (TG) level of 1660 mg/dL. The patient was admitted to the emergency department and repeat blood tests confirmed severe hypertriglyceridemia (TG 1225mg/dL, total cholesterol (TC) 261mg/dL, HDL-cholesterol 37mg/dL, LDL-cholesterol 55mg/dL). Her past medical history was notable for sarcoidosis with mediastinal and dermatological involvement, obstetric antiphospholipid syndrome, former tobacco use and untreated dyslipidemia (2023: TG 178mg/dL, TC 222mg/dL, HDL-cholesterol 47mg/dL, LDL-cholesterol 139mg/dL). Her father had a medical history of dyslipidemia, with no other relevant family history. Treatment was initiated with high-dose omega-3 supplementation (4.5g/day) and a low-fat diet tailored by clinical nutrition. The patient remained hospitalized for clinical and laboratory monitoring, showing a progressive decline in triglyceride levels. Outpatient follow-up was maintained, with pre-delivery triglycerides decreasing to 609 mg/dL. At 36 weeks and 5 days, the patient underwent cesarean delivery due to premature rupture of membranes. A healthy female neonate was delivered (2725g; 46.50cm) with Apgar scores of 9/10/10 at 1, 5, and 10 minutes, respectively. Eighteen days after delivery, TG levels decreased significantly (321mg/dL).

Conclusion: A multidisciplinary approach is essential to guide management and prevent severe complications in gestational hypertriglyceridemia. Dietary intervention and pharmacological treatment can effectively lower lipid levels during pregnancy, which typically decline significantly postpartum.

PO04

ACUTE FATTY LIVER OF PREGNANCY WITH INTRAUTERINE FETAL DEATH IN A WOMAN WITH AUTOIMMUNE POLYGLANDULAR SYNDROME TYPE 2 – A CASE REPORT

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Introduction: Pregnant women with autoimmune disorders are at increased risk of adverse maternal and fetal outcomes. Acute fatty liver of pregnancy (aflp) is a rare but potentially life-threatening obstetric emergency, typically occurring in the third trimester and presenting with nonspecific clinical features. We present a rare case of aflp complicated by intrauterine fetal death in a pregnant woman with autoimmune polyglandular syndrome type 2 (aps-2).

Case presentation: A 37-year-old woman with aps-2, including addison's disease and autoimmune thyroiditis with hypothyroidism, diagnosed at age 26, was admitted during her second pregnancy. Her first pregnancy, at age 29, had been complicated by gestational diabetes requiring insulin therapy, with normal postpartum oral glucose tolerance testing. The pregnancy resulted in a healthy newborn with normal anthropometric parameters. At the beginning of her second pregnancy, she was treated with hydrocortisone (10+5 mg/day), fludrocortisone (150 µg/day), and levothyroxine (112 µg/day), with multidisciplinary obstetric-endocrine follow-up initiated at 9 weeks. At 15 weeks, type 1 diabetes mellitus (t1dm) was diagnosed and insulin therapy initiated. Throughout pregnancy, adrenal and thyroid function remained adequately controlled, with satisfactory glycemic control. Blood pressure remained normal, and fetal ultrasounds and echocardiography were unremarkable. At 29+6 weeks of gestation, she presented with vomiting, peripheral and abdominal edema, headache, generalized abdominal pain, and recurrent hypoglycaemia over the preceding 4 days, with normal fetal movement perception until admission. She was normotensive and afebrile. Ultrasound revealed fetal asystole, with normal amniotic fluid volume. Laboratory investigations showed marked cytolysis, hyperbilirubinaemia, elevated creatinine and hyponatremia. Aflp was suspected, and an emergency caesarean section was performed. Postoperatively, she required 7 days of intensive care for cardiovascular, respiratory, hepatic, and renal dysfunction and was discharged home after a total hospital stay of 12 days.

Conclusion: this case illustrates a fulminant presentation of aflp complicated by intrauterine fetal death in a woman with aps-2 and t1dm diagnosed during pregnancy, despite close multidisciplinary surveillance. The coexistence of aflp and aps-2 has not been well described and may pose additional challenges in clinical management. Furthermore, manifestations of endocrine decompensation may overlap with the early and nonspecific presentation of aflp, potentially delaying its recognition.

PO05

RISING CALCITONIN IN A TWIN PREGNANCY: MANAGING INCOMPLETE BIOCHEMICAL RESPONSE IN MEN2A

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Introduction: Men2a, caused by germline ret mutations, combines medullary thyroid carcinoma (mtc), pheochromocytoma and primary hyperparathyroidism. In pregnancy, calcitonin, the cornerstone of mtc surveillance, also has placental and mammary origins, while imaging and intervention are constrained.

Case report: A 46-year-old woman underwent total thyroidectomy in 2021 for two nodules (bethesda v and iv); histology showed a 7 mm medullary carcinoma, a type unsuspected preoperatively, so node dissection was not performed. An excellent response (undetectable calcitonin, negative cervical ct) obviated reintervention. Germline testing established men2a (ret c.1831t>g, p.cys611gly, exon 10). Right adrenalectomy for pheochromocytoma followed in 2022, with calcitonin undetectable through 2023. Markers subsequently rose, and ⁶⁸ga-dotatoc pet/ct (december 2025) showed faint uptake in the thyroid bed (suvmax 3.1) and a left cervical node (2.3), indicating low somatostatin-receptor expression. Preconception (february 2026): calcitonin 60.7 pg/ml, cea 16.3 ng/ml, negative metanephrines. She then conceived a dichorionic-diamniotic twin pregnancy via frozen embryo transfer with donor oocytes. At 11 weeks, calcitonin had risen to 481.0 pg/ml and cea to 52.2 ng/ml (doubling time <2 months); ionised calcium and pth excluded hyperparathyroidism. Second-trimester ultrasound showed stable disease (5x5x6 mm thyroid-bed lesion; 7x4x7 mm level va/vb node). Biopsy and ionising restaging were deferred to postpartum. Pregnancy is ongoing uneventfully.

Discussion: Incomplete biochemical response preceded conception, so the rise cannot be attributed to gestation alone. However, extrathyroidal calcitonin production, amplified by a twin placenta, renders the calcitonin doubling time uninterpretable in isolation. Lactation sustains this confounding, so calcitonin should be re-baselined only after weaning. Cea, by contrast, rose in parallel with a similarly short doubling time and has no major gestational source. The concordant rise supports true disease progression rather than a physiological artefact. Although calcitonin >150 pg/ml mandates exclusion of distant metastases, ionising imaging was deferred, leaving ultrasound and non-contrast mri. Syndromic screening continued, using ionised calcium, since albumin-bound calcium falls physiologically in pregnancy. Donor oocytes eliminated ret transmission risk.

Conclusion: In men2a, pregnancy suspends the usual biochemical decision rules for mtc. Structural surveillance continued syndromic screening and planned post-lactation reassessment and restaging preserve oncological safety without compromising gestation

PO06

MAGNESIUM SULPHATE-INDUCED HYPOCALCAEMIA IN PREGNANCY: CASE OF DIAGNOSTIC COMPLEXITY

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Introduction: Eclampsia is defined as new-onset seizures associated with hypertensive disorders of pregnancy after exclusion of alternative neurological, metabolic, or toxic causes. Intravenous magnesium sulphate is the standard anticonvulsant treatment. However, hypermagnesaemia may induce hypocalcaemia through parathyroid hormone suppression and increased urinary calcium loss, potentially creating diagnostic uncertainty when seizures occur in pregnancy.

Case Report: A 37-year-old woman at 28+6 weeks' gestation presented after a 5-minute generalised tonic-clonic seizure, with spontaneous recovery and postictal amnesia. Her history included gestational diabetes, fetal growth restriction in a previous pregnancy, superficial venous thrombosis with heterozygous factor V Leiden mutation, and gestational hypothyroidism. She had no thyroid or cervical surgery, autoimmune disease, or family history of hypocalcaemia. On admission, she was conscious and haemodynamically stable (blood pressure 114/73 mmHg; heart rate 102 bpm). Obstetric assessment was reassuring. She denied perioral or peripheral paraesthesias. Blood tests showed normal platelet count ($152 \times 10^3/\mu\text{L}$), renal and liver function, sodium (133 mmol/L), and mild proteinuria (100 mg/dL). Presumed eclampsia was treated with high dose of intravenous magnesium sulphate and dexamethasone. After 4 hours of magnesium perfusion, subsequent testing showed magnesium of 4.9 mg/dL, corrected calcium of 7.4 mg/dL, and QTc prolongation (483 ms). Brain magnetic resonance imaging with angiography and electroencephalography were unremarkable. Magnesium sulphate was discontinued, and intravenous calcium gluconate was administered, resulting in progressive normalisation of serum calcium. Further investigations showed 25-hydroxyvitamin D of 38 nmol/L and parathyroid hormone of 146 pg/mL. Treatment with calcium carbonate, calcitriol, cholecalciferol normalised calcium, phosphate, and vitamin D concentrations. No further seizures or hypocalcaemic symptoms occurred during admission. Labour was induced at 37 weeks without complications. The patient is currently being studied by neurology.

Conclusion: Although hypocalcaemia may cause seizures, the absence of hypocalcaemic symptoms before presentation and the temporal relationship with magnesium sulphate infusion suggest that treatment-related hypermagnesaemia precipitated clinically significant hypocalcaemia. Calcium and magnesium monitoring should be considered during magnesium sulphate therapy, particularly in patients with elevated magnesium concentrations, electrocardiographic abnormalities, or risk factors for hypocalcaemia.

P07

THE MISSING GENE: A CHALLENGING CASE OF CONGENITAL ADRENAL HYPERPLASIA INTRODUCTION

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Introduction: Congenital adrenal hyperplasia (CAH) encompasses a group of disorders caused by mutations in genes encoding enzymes involved in steroidogenesis. In 95% of cases, it corresponds to mutations in the CYP21A2 gene, causing 21-hydroxylase enzyme deficiency. It can be divided into classic and non-classic forms depending on the severity of the enzymatic deficit. Non-classic forms tend to be milder, presenting later in life (adolescence or adulthood) with hirsutism, ovulatory dysfunction, or infertility, or may even be asymptomatic.

Case Report: A 26-year-old patient, G1P1A0, with an uncomplicated pregnancy in 2016, was referred to an infertility outpatient consultation after two years of unsuccessful attempts to conceive. She presented with regular cycles and menstrual periods lasting 4 to 5 days. Initial workup revealed a 17-OH-progesterone level of 4.29 ng/mL, and she was referred to the endocrinologia department. There, an ACTH stimulation test was performed, with 17-OH-progesterone at 60 minutes post-stimulation of 10.1 ng/mL, compatible with a diagnosis of CAH. Treatment with hydrocortisone was initiated, genetic testing was requested, an abdomino-pelvic MRI was ordered, and a medical genetics consultation was arranged for both the patient and her partner. A multigene panel for congenital adrenal hyperplasia detected no relevant variants, and the MRI showed no adrenal nodular lesions or signs of hyperplasia, nor any ovarian nodular lesions. The patient was evaluated at the genetics consultation, which cleared her for pregnancy without the need for further investigations, and she is currently awaiting medically assisted reproduction.

Conclusion: Non-classic CAH may present as infertility and should be systematically investigated in such cases. This case is notable for the discordance between the biochemical diagnosis and the absence of genetic or radiological confirmation. The borderline value may partly account for the negative multigene panel result, as milder forms may correspond to rare mutations. This case highlights the possibility that some forms of non-classic CAH may not be genetically confirmed even when a biochemical diagnosis has been established.