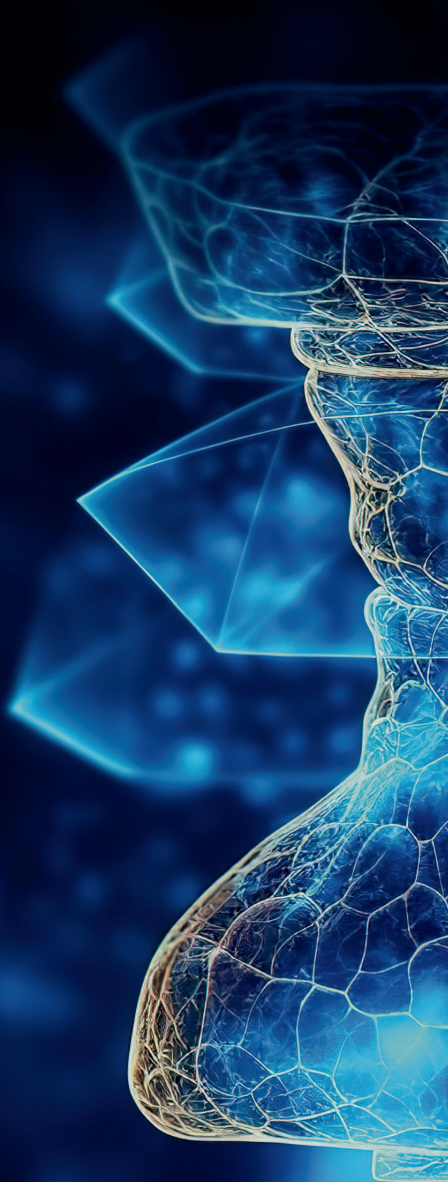


BOOK OF ABSTRACTS:

NEW CHALLENGES IN THE DIAGNOSIS AND TREATMENT OF SELECTED ENDOCRINOPATHIES AND CO-OCCURRING DISEASES IN THE LIGHT OF CURRENT RECOMMENDATIONS



September 25, 2026

Polish Academy of Sciences
Scientific Centre in Paris



Paris
Centre Scientifique



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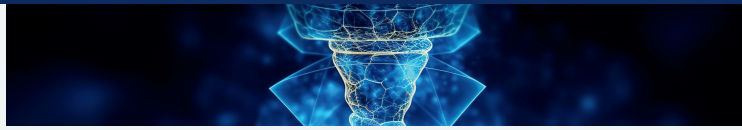
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New challenges in the diagnosis and treatment of selected endocrinopathies and co-occurring diseases in the light of current recommendations

9:00 Opening remarks

- **Prof. Aldona Jankowska** – Polish Academy of Sciences Scientific Centre in Paris, France
- **Prof. Artur Bossowski** – Medical University of Białystok, Poland

I. Diabetology session:

9:10 Prof. Michel Polak, MD, PhD Université Paris Cité, INSERM U1016, Institut IMAGINE, Hôpital Universitaire Necker Enfants Malades, France
Monogenic diabetes beyond the insulin cell: neuropsychology at play

9:35 Prof. Jason Gaglia, Harvard Medical University, USA
Tolerizing Drugs in Type 1 Diabetes: Immune Reprogramming for Beta-Cell Preservation

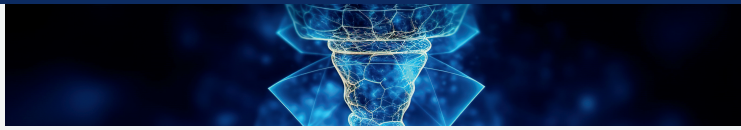
10:00 Prof. Piotr Trzonkowski, MD, DSc, Medical University in Gdańsk, Poland
Type 1 diabetes mellitus – breaking immune tolerance

10:25 Prof. Evelien Gevers, MD, PhD, Royal London Hospital, London, UK
Challenges in the treatment of type 2 diabetes in children

10:50 Coffee break & group photo of lecturers and young scientists (poster session)

II. Thyroid session:

11:20 Prof. Ashlok Bhaseen, President of the Thyroid Federation International, Canada
Current aspects of nutrition in thyroid disease and other endocrinopathies



11:45

Prof. Artur Bossowski, Medical University in Białystok, Poland

Modern diagnostic methods for nodular goiter and thyroid cancer in children and adolescents in light of current recommendations

12:10

Prof. Małgorzata Waśniewska, MD, DSc, School of Medicine, University of Messina, Italy

Lessons learned from 50 years newborn screening for congenital hypothyroidism

12:35

Prof. Jadwiga Furmaniak, MD, DSc, School of Medicine, Cardiff University, UK
Polyendocrine disorders – current perspectives on diagnosis and treatment

13:00

Lunch break & poster session

Moderators:

Prof. Barbara Głowińska-Olszewska, Doc. Hanna Borysewicz-Sańczyk, Dr Beata Sawicka, Medical University in Białystok, Poland

III. Varia:

14:00

Prof. Bart Roep, Medical University in Leiden, Netherlands

Inversing successful anti-cancer immunotherapies to cure autoimmune disease

14:25

Dr. Dulanjalee Kariyawasam, MD, PhD, Paris Cité University, Endocrinology, Diabetology, Gynaecology Department – Necker – Enfants Malades University Hospital, INSERM U1016 – Institut Cochin, France

Metabolic risk in pediatric brain tumors survivors

14:50

Prof. Iwona Beń-Skowronek, MD, DSc, Medical University in Lublin, Poland
Current aspects of diagnosis and treatment of achondroplasia – what we know one year after the launch of the drug programme

15:15

Prof. Mariusz Ratajczak, Louisville University, USA, WUM

Somatotropic signaling and developmental early stem cells in the adult tissues

DIABETOLOGY SESSION



Pr Michel Polak

Endocrinologie, gynécologie et diabétologie pédiatriques, hôpital universitaire Necker—Enfants-Malades, Assistance publique—Hôpitaux de Paris, équipe du réseau de référence européen ENDO-ERN (European Reference Network), du réseau maladie rare PRISIS (Pathologies Rares de l'Insulinosécrétion et de la Sensibilité à l'Insuline), 149, rue de Sèvres, 75015 Paris, France

Université Paris Cité, Paris, France

Inserm U1016, Institut Cochin, Paris, France

Inserm U1163, Institut IMAGINE, Paris, France

Membre de l'Académie Nationale de Médecine de France

Tolerizing Drugs in Type 1 Diabetes: Immune Reprogramming for Beta-Cell Preservation

“Neonatal” diabetes mellitus is defined by the presence of hyperglycemia linked to a lack or insufficiency of circulating insulin. It is a rare genetic disease (occurring in 1 out of every 100,000 live births in France). It typically manifests before the age of six months, and rarely between six months and one year. The disease is associated with two main categories of mechanisms: pancreatic malformation involving impaired development and/or survival of insulin-secreting cells (beta cells), or functional abnormalities in existing pancreatic beta cells—such as mutations in the **ABCC8** and **KCNJ11** genes, which encode the pancreatic beta-cell potassium channel. All patients carrying mutations in the **ABCC8** or **KCNJ11** genes—which are also expressed in the central nervous system—exhibit at least some neurological and/or neuropsychological disorders. These patients can often successfully switch from insulin injections to oral sulfonylureas. It has also recently been shown that sulfonylureas (specifically glibenclamide) can improve neurological, neuropsychological, and visuomotor deficits if administered early in the affected child's life. Elucidating the molecular basis of this rare disease has confirmed the importance of genes encoding proteins involved in the development and function of insulin-producing cells. This genetic insight has also shed light on the role these genes play in the pathogenesis of diabetes mellitus in both children and adults, illustrating the value of studying a rare disease to enhance our understanding of a common one: diabetes mellitus.

Prof. Jason Gaglia
Harvard Medical University, USA

Tolerizing Drugs in Type 1 Diabetes: Immune Reprogramming for Beta-Cell Preservation

Type 1 diabetes (T1D) arises from immune-mediated destruction of insulin-producing pancreatic beta cells, driven by autoreactive T lymphocytes, antigen-presenting cells, inflammatory cytokines, and impaired regulatory immune mechanisms. Although exogenous insulin is essential for metabolic control, it does not address the underlying autoimmune process or preserve endogenous beta-cell mass. Immune-tolerizing therapies aim to alter the disease trajectory by selectively reprogramming pathogenic immunity while preserving protective responses to infection and malignancy. Rather than inducing broad or continuous immunosuppression, the central therapeutic objective is restoration of durable immune regulation directed at beta-cell antigens.

Current clinical and translational interventions span several complementary mechanisms: transient T-cell modulation or depletion; blockade of co-stimulatory and inflammatory pathways; expansion and functional enhancement of regulatory T cells; B-cell-directed therapies; cytokine-pathway inhibition; and antigen-specific approaches using insulin, proinsulin, glutamic acid decarboxylase, or other islet antigens. Anti-CD3 therapy has provided clinical proof of principle that immune intervention can delay progression from presymptomatic autoimmunity to clinical T1D and transiently preserve stimulated C-peptide in selected individuals with recent-onset disease. However, the magnitude and durability of benefit with this therapy remains variable highlighting the need for continued development and alternative approaches.

Prof. Piotr Trzonkowski, MD, DSc

PolTREG S.A., Medical University of Gdańsk, Poland

Type 1 diabetes mellitus – breaking immune tolerance

T regulatory cells (TREGs) are considered a viable option in tolerizing treatment in the clinic. Promising clinical experiments and trials with clinical-grade Tregs cultured as advanced therapy medicinal product (ATMP) are completed already. In our centre, TREGs have been tested in clinical trials in graft versus host disease, type 1 diabetes and neuroinflammatory diseases with up to 12 years of the follow up of the patients. We will present the path from preclinical studies to the results of clinical trials and recent marketing authorisation (MAA) experience with European Medicinal Agency (EMA). It was 12-years-lasting long-term follow up of the patients and comparisons of the patients who responded to the therapy versus non-responders which allowed us to develop Treg products with the most efficient clinical profile. These clinical results were associated with immune profile of the patients which allowed us to build new strategies of the therapy of T1D which are in preclinical stage now. The focus is on autoantigen-specificity and expression of checkpoint receptors in T cell compartment.

It is our hope that our TREG-based therapies will be available in 2027 for routine treatment in Europe upon MAA approval by EMA.

Prof. Evelien Gevers, MD, PhD
Royal London Hospital, London, UK

Challenges in the treatment of type 2 diabetes in children

The incidence of type 2 diabetes in children and young people is increasing globally, following the increase in prevalence of obesity in children and young people.

Type 2 diabetes in children and young people is more severe than T2D in older people. In addition people diagnosed with T2D at young age, will be living with diabetes for a much longer time. Previous work from the TODAY study shows that after 15 years of living with T2D, approx. 50% of people have complications in the form of hypertension, liver disease, renal disease, and nerve disease. Life expectancy is severely reduced in people diagnosed with T2D at young age.

Treatment options for T2D lag behind the options available in adults due to delay in clinical trials in children compared to adults. However, we have now moved on from insulin and metformin as only treatment options to GLP1 receptor agonists and SGLT2 inhibitors. The number of available GLP1RA is still small in children but increasing in adults, showing that newer generation drugs may be more effective. CGM may also play a role in management of T2D. Role and effectiveness of exercise and other non-drug treatments in management of T2D will also be discussed.

Rates of complications in T2D are high, and guidelines for treatment for complications will also be discussed.

Time may tell us if these new approaches in management will improve outcomes.



THYROID SESSION

Ashok Bhaseen, M.Pharm, MMS

President Thyroid Federation International, Toronto, Canada

Current aspects of nutrition in thyroid disease and other endocrinopathies

Thyroid health is closely linked to adequate nutrition, as several essential micronutrients are required for the synthesis, metabolism, and normal functioning of thyroid hormones. This presentation highlights the importance of iron, selenium, vitamin D, iodine, and zinc in maintaining thyroid function and overall health. Iron supports thyroid hormone synthesis and the activity of thyroid peroxidase, while selenium contributes to the conversion of T4 to the active T3 hormone and provides antioxidant protection. Iodine is an essential component of thyroid hormones, and zinc plays an important role in thyroid hormone metabolism and immune function. Vitamin D is also associated with immune regulation and healthy thyroid function.

Dietary sources of these nutrients include seafood and fish, eggs, dairy products, meat, legumes, nuts and seeds, whole grains, leafy green vegetables, and fortified foods. Some foods, such as seafood, eggs, dairy products, and certain nuts and seeds, can provide several of these essential nutrients at the same time. A balanced and varied diet can therefore play an important supportive role in maintaining thyroid health and preventing nutritional deficiencies.

Prof. Artur Bossowski, MD, PhD

Department of Pediatrics, Endocrinology and Diabetes with a Cardiology Unit, Medical University of Białystok, Poland

Modern diagnostic methods for nodular goiter and thyroid cancer in children and adolescents in light of current recommendations

Thyroid cancer is the most common malignant tumor of the endocrine glands in both children and adults. Of the more than 2,500 new cases of thyroid cancer diagnosed each year, approximately 2.0% occur in people under the age of 19. This corresponds to approximately 40–50 new cases per year (1.3 cases per 1,000,000 inhabitants). In girls, this cancer is the second most commonly diagnosed solid tumor (after CNS cancers), and in boys, it is the eighth most commonly diagnosed solid tumor. The incidence of thyroid cancer increases with age; in the 1–14 age group, it is 0.35 per 100,000, while in the 15–29 age group, it rises to 6.08 per 100,000. The etiopathogenesis of focal lesions is not fully understood, although predisposition to neoplastic transformation is influenced by genetic, environmental, and hormonal factors. Factors stimulating thyroid growth include thyrotropin, tissue growth factors (including IGF-1, epidermal growth factor [EGF], fibroblast growth factor [FGF], and platelet-derived growth factor [PDGF]), cytokines, and immunoglobulins, including TGI—an immunoglobulin that stimulates thyroid growth. Additionally, the role of iodine deficiency, exposure to ionizing radiation, autoimmune thyroid diseases, and environmental pollutants is emphasized. Genetic factors play an important role, particularly in the development of papillary thyroid carcinoma—mutations leading to activation of the MAPK (mitogen-activated protein kinase) pathway predominate—RET/PTC rearrangements (approximately 11 types have been described to date), BRAF V600E and TART C228T mutations; in follicular thyroid cancer—PAX8/PPARG rearrangements; and in medullary thyroid cancer—20% of cases are hereditary tumors caused by mutations in the RET proto-oncogene (point mutations). The role of genes is also evidenced by the co-occurrence of thyroid cancer in genetic syndromes such as familial adenomatous polyposis (FAP), Gardner syndrome, Cowden syndrome, DICER1 syndrome, Carney syndrome, and Werner syndrome. The clinical presentation is associated with the presence of a lump or lumps in the neck, enlargement of one or more cervical lymph nodes, vocal cord paralysis (hoarseness, aphonia), shortness of breath, swallowing difficulties,

and, in rare cases, Horner's syndrome. A diagnostic workup algorithm was published in Polish Endocrinology 2024;75(6):565–591 entitled "Diagnostics and Treatment of Differentiated Thyroid Carcinoma in Children—Guidelines of the Polish National Scientific Societies, 2024 Update." It therefore includes analysis of fT3, fT4, and TSH levels, as well as thyroid antibodies (due to the possibility of coexisting autoimmune processes), and, in some cases, molecular testing. Imaging studies may include a chest X-ray, thyroid ultrasound with possible elastography, CT or MRI, and scintigraphy. The gold standard for diagnosis remains ultrasound-guided fine-needle aspiration biopsy (FNA) for focal lesions that exhibit clinical and/or ultrasonographic features indicative of an increased risk of malignancy. The currently accepted system for cytological diagnosis of biopsy specimens is the Bethesda system, developed by the National Cancer Institute (NCI). The mainstay of treatment is surgery, the extent of which depends on the cytological classification, the clinical or ultrasound-based risk of malignancy, the stage of the disease according to the TNM classification, the assessment of the risk of recurrence, and the results of molecular testing. Additionally, in differentiated thyroid cancers, radioactive iodine therapy is used (for patients over 5 years of age) to destroy any remaining thyroid tissue after surgery (residual thyroid ablation). In summary, it should be noted that in children: 1) the incidence of thyroid cancer continues to rise (95% of cases are papillary thyroid carcinoma [PTC]; follicular carcinoma accounts for 5%, while oxophilic and undifferentiated carcinomas are extremely rare), 2) the disease course is multifocal, with more frequent metastases to lymph nodes (78% of cases at the time of diagnosis) and to the lungs (9–30%), 3) the prognosis for children with PTC is very good—better than in adults—and the 10-year survival rate is 98%.

Prof. Małgorzata Waśniewska, MD, DSc
School of Medicine, University of Messina, Italy

Lessons learned from 50 years newborn screening for congenital hypothyroidism

Congenital hypothyroidism (CH) may result from a developmental or functional defect of the thyroid gland (primary CH), or to hypothalamic-pituitary axis dysfunction (central CH), or more rarely from abnormalities in the action, transport, or metabolism of thyroid hormones. Thyroid hormone (TH) is essential for brain development in utero and during the first 2 to 3 years of life. The negative effects of TH deficiency on brain development are irreversible.

In the absence of prompt initiation of replacement therapy during the neonatal period, CH can lead to profound impairment in growth and neuropsychomotor development. Since signs of CH early in life can be subtle and difficult to recognize, early detection and prompt treatment goals have been achieved by newborn screening (NBS), which was started fifty years ago.

Over the past decades, substantial progress has been made in NBS for CH. Enhanced test sensitivity and the broader implementation of re screening strategies for infants at risk of delayed TSH elevation have resulted in a higher number of CH diagnoses compared with the past.

The improvement of NBS and advances in the management of CH have dramatically enhanced patient outcomes through increasingly personalized care from birth to adulthood. Although overall outcomes are generally favourable, ongoing monitoring of treatment adequacy and patient adherence remains essential.

Over the years, it has become increasingly clear that the management of CH requires a comprehensive multidisciplinary approach to ensure optimal long-term health outcomes. Continuity of care is particularly critical during the transition from paediatric to adult services.

Now, 50 years after the introduction of NBS for CH, only a few countries around the world have a screening program capable of detecting central CH.

Prof. Jadwiga Furmaniak, MD, DSc

Honorary Visiting Professor Cardiff University School of Medicine Fellow of The Learned Society of Wales, United Kingdom

Polyendocrine disorders – current perspectives on diagnosis and treatment

The co occurrence of autoimmune diseases was recognised as autoimmune polyglandular syndromes about 65 years ago. It was observed that endocrine disorders are frequently associated with other autoimmune conditions. Today, more than 100 autoimmune diseases have been described, many of which coexist with endocrine diseases, with autoimmune thyroid disorders lying at the centre of many of these clusters. The pathogenetic mechanisms responsible for the co occurrence of autoimmune diseases are not yet fully understood, with the exception of APS 1.

Recently, there has also been progress in the early diagnosis, risk prediction and even prevention of patients with complex endocrinopathies. Notably, advances in hormone replacement therapies have significantly improved patients' quality of life. Novel therapies are emerging in particular in type 1 diabetes and Graves' disease.

At the same time, the broader use of therapeutic antibodies designed to activate the immune system in cancer treatment has revealed that some patients develop undesirable autoimmune conditions, including endocrinopathies.

Furthermore, the latest data on co-morbidities and mortality in autoimmune diseases highlight the need to develop strategies for monitoring and preventing risk factors in order to improve long term patient outcomes.

These advances have led to a significant improvement in quality of life and overall survival for patients with polyendocrine disorders and other autoimmune diseases.



VARIA

Prof. dr. Bart O. Roep

Department of Internal Medicine, Leiden University Medical Center, Leiden, The Netherlands

Inversing cancer immunotherapy to cure autoimmune diseases

Type 1 diabetes mellitus is believed to result from destruction of the insulin-producing β -cells in pancreatic islets that is mediated by autoimmune mechanisms. The classic view is that autoreactive T cells mistakenly destroy healthy ('innocent') β -cells. I propose an alternative view in which the β -cell is the key contributor to the disease. By their nature and function, β -cells are prone to biosynthetic stress with limited measures for self-defence. β -Cell stress provokes an immune attack against the source of a vital hormone. This view would explain why immunotherapy at best delays progression of type 1 diabetes mellitus and points to opportunities to use therapies that revitalize β -cells to reverse the disease, in combination with immune intervention strategies that preferably negotiate with immune system rather than suppress it. The latter could lead to infection and cancer. Conversely, anti-tumor immunotherapy activating the immune system can cause autoimmune diabetes. Hence cancer and type 1 diabetes are opposites, which may imply that inversing successful cancer immunotherapies may offer novel strategies to successfully halt disease progression in type 1 diabetes. I will present proof of concept that this indeed is the case and demonstrate that cancer offers a blueprint to guide studies in type 1 diabetes on disease heterogeneity, personalized medicine, auto (anti-tumor) immunity and immunotherapy.

Prof. Iwona Beń-Skowronek

Department of Paediatric Endocrinology and Diabetology with the Endocrine-Metabolic Laboratory, Medical University of Lublin, Poland

Current aspects of diagnosis and treatment of achondroplasia – what we know one year after the launch of the drug programme

Authors:

Iwona Ben-Skowronek¹, Wiktoria Wróbel^{1,2}

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Coauthors:

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Background:

Achondroplasia is a genetic disorder classified among skeletal dysplasias, leading not only to significant growth impairment and disproportionate body stature but also, due to disturbed endochondral ossification, to numerous medical complications, including orthopedic, otolaryngological, neurological, and even life-threatening respiratory issues. The introduction of causal treatment with vosoritide within a national drug program for children in Poland represents a significant step toward improving the standard of care for this patient population.

Methods:

The study was based on anonymized data from pediatric patients with achondroplasia. Data were collected using structured questionnaires completed by treating physicians from 17 reference centers across Poland, including rare disease, endocrinology, and orthopedic units. All participating centers met the formal requirements for the implementation and management of the national drug program for achondroplasia in Poland.

Results:

Prior to the implementation of the national drug program, access to treatment was limited to a small, highly selected group of patients (approximately a dozen children nationwide). Following program initiation and state reimbursement, the number of treated patients increased substantially to 216 children with achondroplasia currently receiving therapy in Poland. Importantly, 100% of enrolled patients underwent a comprehensive and standardized diagnostic evaluation, including assessment of growth velocity, confirmatory genetic testing, MRI of the central nervous system with detailed assessment of the foramen magnum, and otolaryngological consultation with formal hearing assessment. After 1 year of treatment patients improved body height mean from $-3,8$ SDS to $-3,1$ SDS, growth velocity was improved. Among treated children no serious adverse events were observed. The program enabled nationwide standardization of both qualification and monitoring protocols, ensuring consistent clinical oversight and improved treatment safety. Collaborative efforts between medical specialists, patient organizations, and the active engagement of parents of children with achondroplasia have been crucial in shaping the therapeutic program, fostering public awareness, and facilitating dialogue with governmental authorities. Such collaboration supports integration the patients and

families, strengthens the sense of support, and enables the exchange of experiences.

Conclusions:

In conclusion, the implementation of vosoritide treatment for achondroplasia within a national drug program in Poland has ensured equitable access to healthcare services and appropriate diagnostic procedures for affected children, with the potential to significantly improve final height outcomes and overall quality of life for patients.

Prof. Mariusz Z. Ratajczak MD, PhD, Dsci, Dhc

Laboratory of Regenerative Medicine, Warsaw Medical University, Warsaw, Poland

Stem Cell Program at JGB Cancer Center University of Louisville, Ky 40202, USA

Somatotropic signaling and developmental early stem cells in the adult tissues

Regenerative medicine is searching for stem cells with the potential to differentiate into all three germ layers. Evidence accumulated that adult tissues harbor a population of very rare stem cells endowed with broad differentiation potential. These dormant, cells described as very small embryonic-like stem cells (VSELs) display several epiblast/germline markers what suggest their embryonic origin and developmental deposition in adult tissues. Recently, we found that VSELs do express several sex hormone (SexHs) receptors and respond *in vivo* to SexHs stimulation. Moreover, since VSELs share several markers characteristic of migrating primordial germ cells (PGCs) and can be specified into long-term hematopoietic stem cells (LT-HSCs) and mesenchymal stem cells (MSCs), this observation sheds new light on the BM stem cell hierarchy. Nevertheless, in spite of the expression of pluripotent stem cell markers, changes in the epigenetic signature of imprinted genes (e.g., by erasure of imprinting at the Igf-2-H19 locus) in VSELs are involved in their resistance to Igf-1/Igf-2 signaling and keep these cells in adult tissues in quiescent state. As reported in several emergency situations related to organ damage (e.g., heart infarct, stroke, skin burns), VSELs can be activated and mobilized into peripheral blood (PB) and in appropriate animal models contribute to tissue organ/regeneration. Our data indicate that mobilization of VSELs into PB is regulated by purinergic signaling, Nlrp3 inflammasome and activation of innate immunity. Interestingly, their number correlates with life span in mice and we noticed a positive effect of regular physical exercise and calorie restriction on ameliorating age-dependent depletion of VSELs from adult tissues. Recently, to bring these cells for potential clinical applications we developed an efficient *ex vivo* expansion strategy for these cells.

POSTER SESSION



This photo was taken during the "Current challenges in the diagnosis and treatment of selected endocrinopathies" conference, which was organised on 20 September 2024 at the Polish Academy of Sciences Scientific Centre in Paris.

Poster presented by:

Magdalena Bossowska

Department of Endocrinology, Diabetology and Internal Medicine, Medical University of Bialystok, Bialystok, Poland

Quantile-based modelling uncovers phenotype-tail-restricted PPARG effects on glycaemia and sex-specific TMEM18 associations with central adiposity

Magdalena Bossowska¹, Filip Bossowski¹, Witold Bauer², Edyta Adamska-Patruno³, Katarzyna Maliszewska¹, Adam Krętowski¹

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³Clinical Research Support Centre, Medical University of Bialystok, Bialystok, Poland

Background and aims:

Association testing anchored to the conditional mean may fail to capture genetic effects that are localised to the extremes of a phenotypic distribution. Applying quantile regression in parallel with standard regression frameworks, we examined whether PPARG rs1801282 (Pro12Ala) and two TMEM18 variants (rs6548238, rs7561317) exert heterogeneous effects across the distribution of OGTT-derived physiological indices, adiposity measures, and metabolic syndrome susceptibility.

Materials and methods:

A total of 1,549 participants were characterised (55% women; median age 43.9 years; 31.9% with obesity; 20.7% meeting metabolic syndrome criteria). Fifteen metabolic traits and nine OGTT-derived parameters — among them the Matsuda index, disposition index, glucose and insulin areas under the curve, and the hepatic insulin resistance index — were evaluated under additive, dominant, and recessive inheritance models with adjustment for age and sex. Quantile regression was fitted at five quantiles ($\tau = 0.10$ – 0.90). Supplementary analyses comprised sex-stratified modelling, assessment of PPARG $\times$ TMEM18 interaction, construction of a weighted genetic risk score (GRS), and estimation of population-attributable fractions.

Results:

In conventional models, carriers of the PPARG rs1801282 G allele exhibited a modest reduction in fasting glucose ($\beta = -2.46$ mg/dL; $p = 0.009$). Quantile regression localised this signal to the upper tail: no association was discernible at the median ($\tau = 0.50$; $p = 0.487$), whereas significant effects emerged at the 75th ($\beta = -3.63$; $p = 0.0004$) and 90th percentiles ($\beta = -4.19$; $p = 0.016$); an equivalent gradient characterised 2-hour glucose ($\tau = 0.90$: $\beta = -12.4$; $p = 0.020$). The corresponding preventive fraction for impaired glucose tolerance reached 33.4% (OR = 0.44; $p = 0.013$). Carriage of the TMEM18 rs6548238 T allele conferred reduced odds of metabolic syndrome (OR = 0.53; $p = 0.007$; preventive fraction 21.7%), yet its influence on central adiposity diverged by sex, proving protective in women (waist circumference $\beta = -3.27$; $p = 0.043$) but unfavourable in male T/T homozygotes (waist circumference +11.6 cm, $p = 0.010$; WHR +0.056, $p = 0.011$). TMEM18 rs7561317 was related to greater whole-body insulin sensitivity (Matsuda index $\beta = +0.51$; $p = 0.037$) and to a lower hepatic insulin resistance index ($\beta = -627$ K; $p = 0.010$). Evidence of PPARG $\times$ TMEM18 epistasis was confined to HDL cholesterol ($\beta = +5.03$; $p = 0.019$). No individual variant withstood FDR correction; the weighted GRS, by contrast, retained significance after adjustment for insulin AUC ($q = 0.014$), fasting glucose ($q = 0.015$), and the Matsuda index ($q = 0.021$).

Conclusions:

The glucose-lowering effect of PPARG Pro12Ala appears restricted to individuals occupying the upper portion of the glycaemic distribution, an observation that argues for risk-stratified rather than indiscriminate genotyping. TMEM18 variants display sex-dependent associations with adiposity alongside an overall protective signal for metabolic syndrome. Taken together, quantile regression and polygenic scoring expose distributional and cumulative genetic effects that remain concealed in mean-based analyses.

Poster presented by:

Magdalena Bossowska

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Presymptomatic type 1 diabetes (T1D) among children from North-Eastern Poland – general population screening

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Introduction:

Type 1 diabetes (T1D) is preceded by a presymptomatic phase in which islet autoantibodies (IAbs) are already detectable and the disease can be staged before clinical symptoms occur. Because most children who develop T1D have no affected first-degree relative, screening limited to at-risk families identifies only a small proportion of future cases. Detection of islet autoimmunity in an unselected population allows metabolic monitoring, education of families and reduction of the risk of diabetic ketoacidosis (DKA) at diagnosis, yet data on its prevalence in Polish children remain scarce. The aim of this study was to assess the prevalence of islet autoantibodies and presymptomatic T1D among children aged 1–9 years from north-eastern Poland.

Materials and methods:

Venous blood samples were obtained from 3,575 children between 1 and 9 years of age and examined using a sequential testing approach. The initial screening included the 3 Screen Islet Cell Autoantibody ELISA together with additional IAA ELISA. Positive result in either of these assays underwent confirmatory testing to determine the presence of specific islet

In autoantibodies. Children with confirmed islet autoimmunity were subsequently invited to attend a follow-up visit, which involved further laboratory investigations, metabolic staging, and educational support.

Results:

Detailed antibody analysis confirmed the presence of islet cell autoimmunity in 7.78% of the children who underwent further testing, while 1.17% of the entire study population had multiple positive islet autoantibodies (IAbs). Among the detected autoantibodies, insulin autoantibodies (IAAs) were the most common, accounting for 50.75% of cases, whereas IA-2 autoantibodies (IA-2As) were the least frequently identified (8.4%). IAAs occurred significantly more often in children with a family history of type 1 diabetes (T1D) ($p = 0.04$) and in those aged 1–3 years ($p = 0.02$). In contrast, the prevalence of IA-2As and ZnT8 autoantibodies (ZnT8As) increased with age ($p = 0.035$ and $p = 0.02$, respectively), while ZnT8As were detected more frequently in girls ($p = 0.024$). Multiple positive islet autoantibodies were observed nearly 2.5 times more often in children with a family history of T1D than in those without such a history. A total of 113 children participated in the follow-up assessment. Among participants with dysglycaemia, approximately 67% had a single positive autoantibody, and two children (1.77%) were diagnosed with Stage 3 type 1 diabetes, both before the development of diabetic ketoacidosis (DKA).

Conclusions:

Results confirm the importance of screening for IAbs in general paediatric population as a method of prediction of T1D development. The study also shows that patients with a single positive islet autoantibody may have already developed dysglycaemia and therefore need monitoring.

Poster presented by:

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Brown adipose tissue status conditions the inter-individual heterogeneity of circulating protein and miRNA responses to acute cold exposure

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*equally contributed to the study as senior authors

Background:

Recruitment of brown adipose tissue (BAT) is under active consideration as a treatment avenue in metabolic disease, yet how BAT engagement is reflected jointly across the circulating proteome and miRNome has received little attention. We asked whether BAT status conditions the multi-omic response elicited by acute cold stimulation.

Methods:

Twenty-five healthy, non-smoking White men completed a standardised 120-minute cooling protocol. BAT activity was quantified by ¹⁸F-FDG PET/MR imaging, and participants were assigned to a BAT(+) (n = 17) or BAT(–) (n = 8) stratum according to a glucose uptake cut-off of >2.0 μmol/(100 g)^{–1}·min^{–1}. Plasma was drawn at baseline and again after 60 and 120 minutes. Two platforms were applied in parallel: a targeted proteomic panel covering 582 human plasma proteins (n = 25), and a NanoString nCounter multiplex assay interrogating 798 endogenous miRNAs (23 participants with complete data following outlier exclusion and background correction).

Results:

Baseline comparison yielded no protein that differed between strata once false discovery rate (FDR) correction was applied, indicating that resting plasma carries no signature

attributable to BAT status. Separation between groups emerged only longitudinally, as BAT $\times$ Time divergence over the cooling period. The most pronounced interaction was carried by vinculin (interaction FDR = 0.076), which rose exclusively in BAT(+) participants ($\log_2\text{FC} \approx +5.6$) while falling in the BAT(−) stratum ($\log_2\text{FC} \approx -8.0$). Recasting the miRNA data as within-subject deltas suppressed the dominant baseline variance component and exposed a transient rise in the composite miRNA activation score at 60 minutes (Cohen's $d = 0.64$; repeated-measures ANOVA $P = 0.030$), an increase evident in 77% of participants. Among individual probes, the clearest effect was sustained downregulation of the pro-inflammatory miR-21-5p at 120 minutes ($d = -0.58$; $P = 0.013$). Global miRNA responsiveness spanned an 8-fold range across subjects, with the separation between high and low responders resting on large effect sizes at particular probes, most notably miR-16-5p (21-fold difference; $P = 0.0019$).

Conclusions:

What distinguishes BAT-positive individuals is therefore a dynamic multi-omic reaction to thermogenic challenge rather than a fixed resting state. The divergent behaviour of vinculin points to a circulating readout of acute cytoskeletal remodelling, while the persistent fall in miR-21-5p is consistent with a concurrent anti-inflammatory metabolic shift. Translationally, following plasma trajectories of mechanotransduction proteins and stratifying patients by miRNA responder phenotype could displace reliance on radioactive imaging, providing the basis for a liquid biopsy framework for monitoring BAT-directed pharmacotherapy in metabolic disease.

Poster presented by:

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Plasma Metabolomic Signature of Pediatric Graves' Disease and Its Association with Circulating Regulatory B Cells and Dendritic Cell Subsets

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Background:

Graves' disease (GD) is an autoimmune thyroid disorder characterized by dysregulated immune responses and altered thyroid hormone production. Although alterations in immune cell populations have been described in pediatric GD, the systemic metabolic environment accompanying these immune changes remains poorly defined. This study aimed to characterize the plasma metabolomic profile of GD patients and to investigate associations between circulating metabolites and selected immune cell subsets.

Methods:

Plasma metabolomic profiling was performed in pediatric Graves' disease patients and healthy controls. Multivariate and univariate statistical analyses were used to identify metabolites differing between groups. Metabolite set enrichment and pathway topology analyses were conducted to identify metabolic pathways represented among altered metabolites. Correlation analyses were subsequently performed between plasma metabolite levels and selected regulatory B cell (Breg) and dendritic cell (DC) phenotypes quantified in peripheral blood mononuclear cells obtained from the same individuals.

Results:

Multivariate analysis demonstrated clear separation between Graves' disease patients and

healthy controls, indicating distinct plasma metabolomic profiles. Most significantly altered metabolites showed reduced concentrations in Graves' disease plasma and were predominantly lipid-related species, including multiple phosphatidylcholines, lysophosphatidylcholines, and sphingomyelins. In contrast, a smaller group of metabolites was increased in Graves' disease, including glutamine, asymmetric dimethylarginine, serotonin, aminoadipic acid, and hydroxyproline. Pathway enrichment and topology analyses indicated that altered metabolites were represented in pathways related to nucleotide metabolism (purine and pyrimidine metabolism), nitrogen-related metabolic processes, lipid metabolism (including sphingolipid metabolism), and glycan-associated biosynthetic pathways, including mannose-type O-glycan biosynthesis and glycosaminoglycan biosynthesis. Correlation analyses revealed associations between several plasma metabolites and circulating immune cell populations, including dendritic cell subsets (CD1c⁺ mDC1, CD141⁺ mDC2, CD303⁺ pDC) and regulatory B cell populations characterized by IL-10 or Foxp3 expression. Several lipid metabolites that were reduced in Graves' disease also demonstrated correlations with these immune cell subsets, with distinct correlation patterns observed between dendritic and regulatory B cells.

Conclusions:

Pediatric Graves' disease is associated with a distinct plasma metabolomic signature characterized predominantly by reduced phospholipid-related metabolites. Associations between specific metabolites and circulating immune cell subsets suggest potential links between systemic metabolic alterations and immune cell phenotypes.

Poster presented by:

Dominika KosiŃo

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46,XY gonadal dysgenesis complicated by gonadoblastoma - case report

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Keywords: gonadoblastoma, gonadectomy, growth disorders, gonadal dysgenesis, menstrual disorders

Gonadoblastoma is a rare tumor that develops almost exclusively in dysgenetic gonads, particularly in patients with Y chromosome material (especially the SRY gene). It is a mixed neoplasm composed of germ cells and sex cord-stromal cells resembling Sertoli or granulosa cells. The highest risk is observed in patients with Swyer syndrome (46,XY complete gonadal dysgenesis), mixed gonadal dysgenesis, and 45,X/46,XY mosaicism. Although gonadoblastoma itself has a low malignant potential, approximately 50–60% of cases are associated with or progress to dysgerminoma, the most common malignant germ cell tumor. Therefore, prophylactic bilateral gonadectomy is recommended in patients with gonadal dysgenesis and the presence of Y chromosome material. We present case of gonadal dysgenesis complicated by the development of a gonadoblastoma from the Department of Pediatrics, Endocrinology and Diabetology with the Cardiology Divisions of the Medical University of Białystok.

Case:

A 13-year-old girl with a 46,XY karyotype was evaluated for tall stature (>97th percentile), clitoromegaly, and delayed puberty. Pelvic ultrasonography revealed the presence of a uterus, while the ovaries were not visualized. Laboratory evaluation demonstrated elevated gonadotropin and testosterone levels, as well as elevated serum β -human chorionic

gonadotropin (β -hCG). Serum levels of 17-hydroxyprogesterone (17-OHP), dehydroepiandrosterone sulfate (DHEA-S), estradiol (E2), growth hormone (GH), and insulin-like growth factor 1 (IGF-1) were within the normal range. Due to the diagnosis of gonadal dysgenesis, the patient underwent bilateral gonadectomy. Histopathological examination of the surgical specimen revealed gonadoblastoma of the right gonad. Combined hormone replacement therapy with estrogen and progesterone was initiated. The patient remains under regular follow-up in the endocrinology and oncology outpatient clinics.

Conclusions:

46,XY gonadal dysgenesis is a rare condition associated with a significant risk of gonadoblastoma. Early recognition of clinical features, genetic assessment, and prophylactic gonadectomy play a key role in preventing malignant complications. Lifelong multidisciplinary follow-up is essential for the management of affected patients.

Poster presented by:

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Identification of a cut-off value of the Single Point Insulin Sensitivity Estimator (SPISE) in a cohort of children and adolescents with obesity

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Background and Objectives:

The Single Point Insulin Sensitivity Estimator (SPISE) index is a surrogate marker of insulin resistance (IR) that does not require insulin measurement, thereby avoiding associated costs and variability. This study aimed to determine an optimal SPISE cut-off for identifying IR in a cohort of children and adolescents with overweight (OW) and obesity (OB).

Methods:

A cross-sectional study was conducted in 150 children and adolescents (mean age 11.59 ± 2.38 years) with OW/OB (mean BMI SDS: 3.23 ± 1.0). Inclusion criteria were age between 2 and 18 years and availability of a complete glyco-lipid profile. Exclusion criteria included genetic or endocrine causes of obesity, diabetes mellitus, acute or chronic illnesses, ongoing pharmacological treatments, and use of drugs affecting glucose metabolism. SPISE was calculated as: $600 \times \text{HDL}^{0.185} / (\text{TG}^{0.2} \times \text{BMI}^{1.338})$. SPISE performance was evaluated against established IR indices (HOMA-IR, QUICKI, TyG index, and TG/HDL-C ratio). Receiver operating characteristic (ROC) curve analysis was used to identify the optimal SPISE cut-off and assess diagnostic accuracy.

Results:

The mean SPISE value was 5.41 ± 1.26 and it was significantly higher in prepubertal compared with pubertal subjects (5.69 ± 1.15 vs 5.23 ± 1.29 ; $p = 0.026$), with no sex-related

differences. SPISE showed significant correlations with HOMA-IR ($r = -0.432$; $p=0.001$), QUICKI ($r = 0.422$; $p=0.001$), BMI ($r = -0.86$; $p=0.001$), BMI SDS ($r = -0.445$; $p=0.001$), waist circumference ($r = -0.647$; $p=0.001$), fasting glucose ($r = -0.172$; $p=0.036$), fasting insulin ($r = -0.428$; $p=0.001$), 120-min post-OGTT insulin ($r = -0.337$; $p=0.001$), uric acid ($r = -0.283$; $p=0.007$), ALT ($r = -0.172$; $p=0.035$), and hepatic steatosis ($r = -0.325$; $p=0.004$). ROC analysis identified a SPISE cut-off of ≤ 5.72 as optimal for detecting IR, with good accuracy (AUC 0.78 vs HOMA-IR; AUC 0.83 vs QUICKI) and high specificity (94.1% and 100%, respectively).

Conclusions:

A SPISE cut-off of ≤ 5.72 represents a reliable and accurate threshold for identifying IR in pediatric patients with OW/OB, independent of insulin measurement. SPISE may serve as a cost-effective and accessible tool for metabolic screening, bridging the gap between clinical anthropometry and laboratory-based assessment of insulin sensitivity.

Poster presented by:

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Dramatic Structural and Biochemical Response of a Pediatric Giant Prolactinoma to Cabergoline Therapy

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Background:

While prolactinomas are rare in pediatric populations, they often reach substantial dimensions before proper identification due to the subtle nature of initial endocrine disruptions. Consequently, adolescent patients frequently present with pronounced mass effects and secondary pituitary deficiencies by the time a diagnosis is made.

Case report:

We present the case of a 16-year-old female patient referred for the assessment of primary amenorrhea and marked hyperprolactinemia (1272 ng/mL). The clinical history revealed a two-year progressive decline in visual acuity and unilateral right-sided hearing loss, notably without galactorrhea. Comprehensive hormonal assays indicated multi-axis suppression, including central hypothyroidism, secondary adrenal insufficiency, and a profoundly suppressed gonadal axis.

Baseline magnetic resonance imaging (MRI) in November 2025 demonstrated a massive (28 x 21 x 31 mm) sellar and suprasellar lesion with cystic features. This mass caused severe compression of the optic chiasm and displaced the pituitary infundibulum, classified as Knosp grade 2. Conservative management was elected, utilizing cabergoline alongside essential hormonal substitution with hydrocortisone and levothyroxine.

The patient exhibited an outstanding initial biochemical response; serum prolactin plummeted to 60.8 ng/mL within the first month and achieved full normalization

(16.6 ng/mL) by the third month of treatment. A recent follow-up MRI conducted in July 2026 confirmed substantial structural regression of the adenoma, with dimensions decreasing to 22 x 13 x 22 mm. Crucially, this shrinkage resulted in the complete decompression of the optic chiasm.

Conclusions:

This case underscores that despite extensive diagnostic delays leading to a massive tumor burden and visual compromise, pediatric macroprolactinomas can be managed highly effectively with conservative dopamine agonist therapy. Cabergoline treatment not only swiftly restored biochemical balance but also provided critical structural regression of the tumor, successfully averting the morbidities associated with neurosurgical interventions.

Poster presented by:

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Psychological burden in adolescents with type 1 diabetes: a single-center assessment and implications for integrated psychodiabetes care

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Keywords: type 1 diabetes, adolescents, diabetes distress, diabetes burnout, fear of hypoglycemia, psychodiabetes, integrated care

Introduction and aim:

Adolescence is a particularly vulnerable stage of life for patients with type 1 diabetes (T1D). The demands associated with continuous glucose monitoring and insulin therapy may lead to substantial psychological burden and hinder effective self-management of the disease. The aim of this study was to assess the prevalence of diabetes-related psychological difficulties and selected mental health symptoms among adolescents with T1D, as well as to identify available forms of psychodiabetes support in the Małopolska Voivodeship.

Materials and methods:

A cross-sectional assessment was conducted among 44 patients with T1D who had been living with the disease for at least one year and were recruited during hospitalization or follow-up visits at the University Children's Hospital in Kraków. Depending on age, participants completed validated screening instruments assessing diabetes distress, diabetes burnout, fear of hypoglycemia, sleep quality, depressive symptoms, anxiety, and stress. The study included, among others, the Diabetes Distress Scale, diabetes burnout questionnaires, a fear of hypoglycemia questionnaire, the Pittsburgh Sleep Quality Index, the Quick Inventory of Depressive Symptomatology, and the DASS-21. In parallel, available psychological, psychotherapeutic, and psychiatric support services for children and

adolescents in the Małopolska Voivodeship were identified and mapped.

Results:

Fear of hypoglycemia was the most prevalent psychological problem, affecting 75% of the patients assessed. Diabetes distress was identified in 69.6% of participants, while diabetes burnout was present in 41%. Approximately 40% of patients had reduced sleep quality. At the same time, the majority of participants did not exhibit clinically significant depressive symptoms on screening. In the DASS-21 assessment, anxiety symptoms were more pronounced than depressive symptoms and stress. The findings suggest that the primary psychological burden in adolescents with T1D may be associated mainly with the daily demands and perceived risks of diabetes management rather than solely with depressive symptoms. The analysis of available services demonstrated the presence of facilities offering psychological, psychotherapeutic, and psychiatric support; however, substantial differences in accessibility and long waiting times for some forms of care were identified.

Conclusions:

Psychological burden is common among adolescents with T1D, with fear of hypoglycemia, diabetes distress, and diabetes burnout being particularly clinically relevant. Psychological assessment should therefore constitute an integral component of comprehensive diabetes care rather than being limited to patients presenting with overt symptoms of mental disorders. Early identification of diabetes-related distress and burnout, combined with timely access to specialized psychological and psychotherapeutic support, may facilitate more effective self-management and improve the quality of life of young people living with T1D. The development of regional psychodiabetes care pathways may help translate the results of psychological screening into effective clinical interventions.

Poster presented by:

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Usefulness of continuous glucose monitoring in pediatric patients with preclinical stages of type 1 diabetes

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Introduction:

With an increasing availability of screening programmes for type 1 diabetes (T1D) in pediatric population, there is a growing need for reliable methods of further monitoring of patients diagnosed with preclinical stages of the disease, in order to detect dysglycemia as early as possible.

Aim:

The aim of the study was to assess the usefulness and effectiveness of continuous glucose monitoring system in children with preclinical stages of T1D.

Material:

43 patients aged 4-15.5 years (58.1% boys) with confirmed presence of islet autoantibodies (IAbs), remaining under the care of Diabetes Outpatient Clinic were included. Among the patients, 18.6% presented 1 positive IAb, 55.8% presented 2 positive IAbs, 18.6% presented 3 positive IAbs, and 7% presented 4 positive IAbs. In metabolic staging, a single positive IAb was determined in 11.6%, stage 1 preclinical T1D in 62.8%, stage 2 preclinical T1D in 25.6%.

Methods:

Patients began monitoring their blood glucose using Freestyle Libre 2 sensors immediately after the initial metabolic staging (65.2%) due to ≥ 2 positive IAbs and/or dysglycemia in OGTT, or later during follow-up (34.8%) due to increasing HbA1c levels in follow-up tests.

Before system initiation, patients and their parents were trained in sensor insertion and use, and interpretation of obtained results. Sensor use was recommended to be used approximately every three months, with HbA1c assessed every three to six months.

Results:

The mean follow-up time was 304 days (± 108). During follow-up, 5 patients (11.6%) progressed to the next stage of the disease based on control OGTT or HbA1c elevation. Significant correlations were observed between: mean sensor glucose and HbA1c at baseline, TAR at baseline and maximum glucose during OGTT, and CV and time above 140 mg% at baseline with maximum glucose and 120-minute glucose at baseline OGTT (all $p < 0.05$). TAR at 6 months of follow-up correlated significantly with HbA1c values observed at 12 and 18 months ($p < 0.05$). Time above 140 mg% at 6 months correlated with HbA1c at 12 months ($p < 0.05$). In the group initially diagnosed with stage 2, TAR at baseline correlated with TAR, time above 140 mg%, and CV at 6 months; and mean glycemia at 6 months correlated with HbA1c at 1 year (all $p < 0.05$). In the group initially diagnosed with stage 1, time above 140 mg% at 6 months correlated with time above 140mg% at 12 months.

Conclusions:

Continuous glucose monitoring can provide significant support in monitoring patients with preclinical stages of T1D, providing useful additional information to help predict further disease progression.

Poster presented by:

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Precocious Puberty as the Initial Presentation of Congenital Adrenal Hyperplasia Due to 21-Hydroxylase Deficiency in a 2-Year-Old Boy: A Case Report

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Keywords: congenital adrenal hyperplasia, 21-hydroxylase deficiency, precocious puberty, androgen excess, premature pubarche

Congenital adrenal hyperplasia (CAH) due to 21-hydroxylase deficiency is characterized by impaired cortisol biosynthesis and excessive adrenal androgen production. In boys without salt wasting, diagnosis may be delayed because external genitalia are typically normal at birth. Progressive androgen excess may subsequently manifest as accelerated linear growth, penile enlargement, premature pubarche, and advanced skeletal maturation.

Case presentation:

A 2-year- and 9-month-old boy was referred to our Clinic for evaluation of precocious puberty. Penile enlargement had been observed since approximately 2 years of age, pubic hair had developed during the preceding 3 months, and accelerated growth velocity had been noted over the previous year. Physical examination revealed height above the 97th percentile, penile length of 6 cm, Tanner stage III pubic hair, and prepubertal testicular volume of approximately 1 mL bilaterally. Bone age was markedly advanced to 9 years. Abdominal ultrasonography demonstrated mildly enlarged adrenal glands, whereas testicular ultrasonography and brain magnetic resonance imaging were unremarkable.

Biochemical evaluation revealed markedly elevated 17-hydroxyprogesterone (17-OHP; 110 ng/mL; reference range 0.1–2.3), androstenedione (20 ng/mL; 0.4–2.6), and

ACTH (122 pg/mL; 10–60). Serum testosterone was elevated for age, while β -hCG was undetectable. The LHRH stimulation test showed an LH/FSH ratio of 0.85. DHEA was within the reference range. Morning and evening cortisol concentrations were 326 and 113 nmol/L, respectively. Aldosterone and serum sodium and potassium concentrations were normal, with no biochemical evidence of salt wasting. These findings supported the diagnosis of CAH due to 21-hydroxylase deficiency.

Oral hydrocortisone treatment was initiated at 7.5–5–5 mg/day and subsequently increased to 10–7.5–5 mg/day. During follow-up, 17-OHP decreased markedly from 110 to 3.1 ng/mL.

Conclusion:

This case highlights 21-hydroxylase deficiency presenting in early childhood with rapidly progressive peripheral precocious puberty without salt wasting. Penile enlargement and premature pubarche accompanied by prepubertal testicular volume, accelerated growth, and markedly advanced bone age should raise suspicion of adrenal androgen excess. Early diagnosis and appropriate glucocorticoid treatment are essential to suppress excessive adrenal androgen production, limit skeletal advancement, and preserve adult height potential.

Poster presented by:

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Clinical Utility of an Ultra-Rapid Turbo Bioassay for Thyrotropin Receptor-Stimulating Autoantibodies in Pediatric Graves' Disease

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Background:

Thyrotropin receptor-stimulating autoantibodies (TSAb) are detected in 95–99% of patients with Graves' disease (GD) and play a central pathogenic role by directly driving thyroid hormone overproduction. As functional biomarkers, TSAb are highly relevant for both diagnosis and disease monitoring. We evaluated TSAb in pediatric patients with different phases of autoimmune thyroid disease (AITD), including GD before and during methimazole therapy, using a novel ultra-rapid bioassay.

Methods:

Serum samples from pediatric patients with AITD and healthy controls were analyzed at the accredited, certified, academic JGU Molecular Thyroid Lab, using the novel Turbo™ TSAb cell-based bioassay (Thyretain®, QuidelOrtho, NJ, USA), with a cyclic AMP readout that correlates with luciferase gene activity. TSAb samples <0.024 IU/L were considered negative. Eighty AITD samples and 35 control samples were included. Median age was 12 and 10.5 years in patient and controls, respectively with a female-to-male ratio of 1.65.

Results:

All healthy control samples were TSAb negative. Among the 80 pediatric samples with AITD at various serological stages, 43 (52.5%) were collected during a hyperthyroid phenotype with pathologically increased serum free thyroid hormone levels, 30 (36.5%) during biochemical hypothyroidism, and 7 (11.0%) during euthyroidism. The Turbo™ cAMP bioassay

detected TSAb in all 36 untreated pediatric GD patients (sensitivity 100%) and in five samples obtained from children on methimazole. Overall, 41 of 80 pediatric AITD samples were TSAb-positive. TSAb correlated with thyroid functional status ($p = 0.028$). Dual TSH-R-Ab positivity (positive TSH-R blocking activity) was identified in three of 80 AITD samples (3.75%).

Conclusions:

This is the largest reported pediatric series of TSAb-positive GD samples assessed using a rapid and reliable TSAb bioassay. TSAb significantly correlated with thyroid functional status, underscoring their biological and clinical relevance. These novel findings support the potential utility of the Turbo™ stimulating bioassay as a valuable tool for the diagnosis and monitoring of pediatric AITD.

Poster presented by:

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Ultrasound screening of the thyroid, parathyroid glands, and cervical lymph nodes in children with type 1 diabetes. Interesting ultrasound images

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Keywords: children thyroid ultrasonography, children thyroid ultrasonography in children with type 1 diabetes, interesting ultrasound images

Type 1 diabetes is an autoimmune disease that requires regular monitoring for prevention, early detection of chronic complications, and coexisting autoimmune diseases. According to Clinical Recommendations on the Management of Individuals with Diabetes – 2026 Position Statement of Diabetes Poland, screening tests (hormonal tests – TSH, fT4, fT3, anti-TPO and anti-TG antibodies, and thyroid ultrasound) should be performed upon diagnosis of type 1 diabetes in children and repeated every two years.

In the Department, in the years 2024-2025, I performed 477 ultrasound examinations of the thyroid, parathyroid glands, and neck lymph nodes in children with type 1 diabetes. Among them, 75 children with type 1 diabetes were diagnosed with autoimmune thyroiditis, 1 child with Graves' disease, 3 children with thyroid cancer (papillary), and 89 children with focal thyroid lesions.

Images of the most interesting and rare cases of ultrasound of the thyroid, parathyroid glands, lymph nodes, and neck organs are presented on the poster.

Case 1:

Images of 3 cases of papillary thyroid cancer, confirmed by cytological examination: a 14-year-old girl with in the right lobe a small hypoechoic lesion sized 3x4x3 mm with a lymph

node metastasis; a 15-year-old girl with in the right lobe hypoechoic lesion growing into a capsule sized 4x5x7 mm; and a 16-year-old boy with in the right lobe isoechoic lesion measuring sized 2x2.5x2.5 mm with a lymph node metastasis.

Case 2:

Ultrasound images in 13-year-old boy with type 1 diabetes was shown to have a three lobed thyroid gland – the two right lobes and one left lobe.

Case 3:

Image a 10-year-old boy has of partial thyroid ectopia located at the foramen cecum of the tongue.

Case 4:

A 12-year-old boy has a double left thyroid lobe.

Case 5:

Image of hypertrophy parathyroid gland in 8-year-old boy with pseudohyperparathyroidism and hypertrophy (dysplastic) parathyroid gland in the mother of a child with pseudohyperparathyroidism.

Case 6:

Image of lateral neck cyst in an 8-year-old girl.

Case 7:

Images of tongue cysts in 9- i 13-years-old girls.

Conclusion:

Screening tests play a crucial role in the early detection of thyroid cancer and other thyroid diseases, most of thyroid diseases detected by ultrasound are asymptomatic. Thyroid focal lesions in children have a higher risk of malignancy than in adults, and even small lesions generate metastases earlier and more frequently. Suspicious focal lesions in children require biopsy. Optimally, thyroid ultrasound screening should be extended to the entire pediatric population.

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