psychological, physical, cognitive, social, functional & behavioral elements. QOL has also become more important in health care practice and research.

Aim of Study assess the QOL in children previously both idiopathic short stature (ISS) and growth hormone deficiency (GHD) were on the list of diagnoses in addition children on treatment with growth hormone (GH) by daily injection.

Methods This cross-sectional study involved 200 children between the ages of 8 and 18 who were diagnosed to have GHD and ISS and were on treatment by GH daily injection at the Pediatric Endocrinology Unit at Minia University Hospital from june 2021 until October 2022.

Results 16% of cases were ISS, and 84% of cases had GHD. Ninety-two percent of our studied cases were already on treatment with recombinant growth hormone (rGH). Children with ISS had significantly reduced physical, psychological, & environmental quality of life. Most affected is the social domain. Comparing treated and untreated groups, there is more emotional affection in the untreated group than the treated group (p less than 0.01).

An evaluation of the various sex classes (boys & girls) in respect to several domains amongst personswas made, but no statistically significant distinction was found.

Conclusion(s) QOL was impaired in cases of short stature, whether on GH theraphy or not. However, our study showed a significant correlation between PedsQL scores for short stature and age if the first administration of treatment donating early GH therapy had better QOL scores.

Delegates' Abstracts

Case Reports

31 UNDERSTANDING GENETIC IMPLICATION IN NEONATAL DM – CASE PRESENTATION

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10.1136/bmjpo-2024-ASPED.31

Background Neonatal diabetes is a monogenic type of diabetes mellitus occurs before the age of 6 months and can be transient or permanent. We present a case of neonatal diabetes with positive mutations in KCNJ11 and HNF4A gene. There is no such case described in literature before. This study aims to highlight the importance of the genetic mutations associated with neonatal diabetes.

Case Presentation Previously healthy, 4-month-old male infant born FT with B Wt of 3.0 kg presented with vomiting. Initial investigation showed BG of 368 mg/dL, bicarb of 8.9 and PH of 7.0. HbA1c 13.4%. Upon DKA was resolved, he was transitioned to SQ insulin and later to insulin pump (omnipod) by 1st week post diagnosis. He had negative T1DM antibodies. At 8 month of life, genetic testing was done. It reports mutation in

1. KCNJ11; KCNJ11-related neonatal diabetes, Autosomal Dominant. c.149 G>A p.(R50Q) Heterozygous AND
2. HNF4A; HNF4A-related disorder, Autosomal Dominant. c.-79 C>T Heterozygous

He was started on Glyburide (Glibenclamide) ~0.2 mg/kg/day in two divided doses. By 3rd day, his basal insulin dose were down to half. By 1st week, he was off insulin with stable blood glucose. His current dose is ~0.03 mg/kg/day at 20 month of age with HbA1c of 5.9%. He gets mild hypoglycemic ~2 hr (in 60's) after the medication dose. Medication effect lasts for ~11 hours. There is no food restriction.

Discussion Mutations in the gene KCNJ11 is among the most common causes of permanent neonatal DM. Mutations in the HNF4A gene can lead to a condition called maturity-onset diabetes of the young type 1 (MODY1), which is a monogenic form of diabetes characterized by early-onset diabetes often before the age of 25. Our case present rare incidence of having both mutations together. As expected, he responded well to Sulfonylureas which closes the KATP channel and initiate insulin secretion which was affected by KCNJ11 mutation. However, the current requirement of Glyburide dose is much lower than expected. It's difficult to say if concurrent HNF4A mutation which can present with hyper-insulinemic hypoglycemia in neonatal period has any implication for low sulfonylurea dose requirement.

Conclusion Genetic diagnosis has significant therapeutic implications in management of neonatal diabetes. Majority of infants with neonatal diabetes due to specific mutations known to respond oral formulation and can be managed without insulin. Timely diagnosis and treatment will result in better glycemic control and reduced morbidity.

32 DIABETES MANAGEMENT NEW TOOLS, CASE SCENARIO

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Background Using CGM data can optimised diabetes care in paediatric type 1 and lead to major change in diabetes self control.

Case Report(s) NORA 13 yrs old girl known case DM type 1 for 6 yrs -Poor control with nephropathy

-Idiopathic SS -height 139 cm (_2.7SD) -GV 4 cm/yr-MPH 150 cm

-Weight 44 kg (-0.7SD)-BMI 19kg/m2 (0.1 SD) Injection site ok

Puberty Stage 3She is on MDI therapy Lantus 16 unit novorapid: 7 U pre breakfast

10 U pre lunch 7–8 U pre dinner TDD =0.9 unit/kg/day

ISF 1:50 Hba1c 15.6% using CGM data to address her problems showed it is mainly behaviour and complain point to strength attaches cgm report in the full details case

Conclusion(s) Using CGM data can optimised diabetes care showed may area you need to work on to enhance diabetes control and not only insulin doses changes but very important part which is behaviour change and motivation aids.

33 COMPLICATED CASE OF DKA

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10.1136/bmjpo-2024-ASPED.33

Background DKA is a severe acute complication of diabetes mellitus.

Thiamine (vitamine B1) is a water-soluble vitamin that plays a key role in aerobic glucose metabolism. It is a cofactor of pyruvate dehydrogenase (PDH), an enzyme that must be

stimulated for entry into Krebs cycle for aerobic metabolism. In thiamine deficit states (as in diabetes - thiamin loss in urine), PHD activity is reduced resulting in a shift in pyruvate metabolism to the anaerobic pathway, increasing lactate production and acidosis.

Case Report(s) We prescribe the case of a 16-month-old baby girl, with insulin-dependent diabetes, who was admitted to the emergency department with tachycardia, altered state of consciousness and severe ketoacidosis.

She has an initial HbA1C at 11% at diagnosis, and the autoimmune workup showed positive level of anti-GAD confirming the autoimmune mechanism of her diabetes mellitus. Her stay in the Intensive Care Unit was unusual and full of complications. She received very high doses of insulin for her age and weight, with a very poor response to insulin and resistant persistent acidosis even after >72 hours of IV insulin therapy.

Metabolic evaluation was done in front of this severe and persistent acidosis, high lactic acid level was found. She was started on large spectrum antibiotics after taking all culture samples, in order to cover an eventual infection. It was difficult to find vein for perfusion, a central line on the femoral vein was performed, but unfortunately 3 days later, she developed a deep and extensive vein thrombosis on this central line. Femoral Vein catheter was then removed urgently, and a Broviac catheter was done with heparin IV administration started urgently.

She started to improve only when she received thiamine intravenous supplement, coenzyme Q10, and L-Carnitin, showing the impact of thiamine as adjunctive therapy in the treatment of DKA and diabetes mellitus.

Mitochondrial disease was then suspected, suspicion of MELAS (mitochondrial encephalopathy, lactic acidosis, stroke like episode), which is a common cause of mitochondrial Diabetes disorders, but unfortunately genetic tests still not realized in this patient because of financial issues.

In conclusion, our patient presented a severe combined diabetic ketoacidosis (DKA) and lactic acid acidosis, showing the importance of understanding the pathophysiological mechanisms underlying lactic acidosis in DKA.

Conclusion(s) Genetic tests must be done in this patient in order to rule out an underlying metabolic or mitochondrial disease, as an uncommon but important cause of diabetes mellitus as endocrinopathy.

This case showed the impact of IV Thiamin administration as an adjunctive therapy for DKA leading to faster resolution of acidosis and improving aerobic metabolism.

34 A PATIENT WITH INSULIN-DEPENDENT DIABETES MELLITUS 1 YEAR AFTER BEING DIAGNOSED WITH MEMBRANOUS GLOMERULONEPHRITIS

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Background Diabetes mellitus is a syndrome of disordered metabolism that lead to inappropriate hyperglycemia which could be basically due to an absolute or relative deficiency of insulin, or a defect in insulin action (insulin resistance). The most common cause in children is type 1 DM which is immune- mediated disease.

T1DM is frequently associated with other autoimmune diseases, most commonly autoimmune thyroid diseases (17–30%), celiac disease and other autoimmune disease such as Addison's disease, autoimmune gastritis and very rarely with rheumatoid arthritis, systemic lupus erythematosus and glomerular disease

Case Report(s) Herein, we report a female patient who initially presented at the age of 9 years to nephrology department with acute renal failure which was proved to be secondary to membranous glomerulonephritis on kidney biopsy.

She was treated with methylprednisolone pulse therapy for 2 weeks and continued on prednisolone 60 mg for 2 months, then tapering over 6 months, and kept on prednisolone 10mg for 1 year according to her clinical situation.

During that period, she received cyclophosphamide monthly for 6 months and started on Mycophenolate mofetil.

1 year after diagnosis she presented with polyuria and polydipsia found to have hyperglycemia and glycosuria with normal VBG and diagnosed with DM type 1 for which started on insulin therapy basal bolus regimen.

Recently (4 months ago) she complained of hair loss, seen by dermatologist and diagnosed with alopecia aerate.

Her thyroid function test is normal, anti TPO ant TG are negative, Celiac screen is negative, Liver enzymes are normal. Her electrolyte, basal cortisol and ACTH are normal

Conclusion(s) Diabetes mellitus type1 could be associated with other autoimmune diseases most commonly Hashimoto's Thyroiditis, and celiac disease.

Our patient was diagnosed with Membranoproliferative glomerulonephritis and later on developed manifestations of other autoimmune diseases (DM type1 and alopecia). It was published in medical literature the association of type 1 Diabetes Mellitus and membranoproliferative glomerulonephritis in young adult, which up to our knowledge this the first case in pediatric patients.

35 LATE-ONSET GLUTARIC ACIDURIA UNVEILED DURING THE DIAGNOSTIC JOURNEY IN A PEDIATRIC PATIENT WITH METABOLIC SYNDROME: EXPLORING THE ASSOCIATION BETWEEN TWO CONDITIONS AND THEIR TREATMENT

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10.1136/bmjpo-2024-ASPED.35

Background We present the case of a 12-year-old female patient with a complex medical history. Initially, she presented with recurrent epistaxis and was subsequently diagnosed with metabolic syndrome. Her diagnostic journey involved high blood glucose levels, autoimmune thyroid disease, obesity, and cardiac abnormalities.

Case Report(s) The patient was diagnosed with diabetes at the age of 10 after experiencing polyuria and polydipsia. Initial lab results indicated high HbA1c and fasting blood glucose levels. Despite the diagnosis of type 2 diabetes and the absence of autoantibodies, her glucose control deteriorated. She was treated with insulin and metformin. Furthermore, autoimmune primary hypothyroidism was discovered, and she was started on levothyroxine. The patient's obesity had been present since early childhood, and her BMI exceeded the 98th percentile. She also exhibited signs of acanthosis nigricans.