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Abstract Book

26-27 October 2024 & 2-3 November 2024

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Comparative Analysis of Arabs vs. Global Practices in Managing Coexisting Type 1 Diabetes and Celiac Disease

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Introduction: Type 1 diabetes (T1D) and celiac disease (CD) are both autoimmune diseases, with CD being a prevalent comorbidity among individuals with T1D for which it is globally recommended to screen people with T1D for CD.

Objectives: This study aims to explore how these coexisting conditions are managed by healthcare providers in Arab countries, comparing their practices with other physicians in the rest of the world.

Methods: A 30-question web-based survey, developed by JENIOUS (ISPAD) and YES (ESPE), was used to gather data on the clinical practices and management strategies employed by physicians treating individuals with both T1D and CD from July to December 2023. A post hoc analysis was conducted to compare responses, highlighting differences between the practice patterns of physicians in Arab countries (Group1) and their peers from other countries (Group2).

For Gluten Free Diet (GFD) initiation when the first CD screening becomes positive; in Group1, 31.8% repeat the tTG-IgA test along with the EmA test, and if values exceed 10 times the upper normal limit (UNL), they start GFD without performing a biopsy. 18.2% perform a biopsy without repeating the test. Whereas in group2, 26.1% follow a similar approach by repeating the tTG-IgA test and starting GFD without a biopsy if values are high. 22.3% prefer to perform a biopsy after repeating the test. 18.8% refer the patient to a gastroenterologist. Therefore, Chi-Square Test for GFD Initiation showed a value of 54.96, p-value: 0.259, (dof):49 indicating no statistically significant difference in the GFD initiation practices between physicians from the two groups.

People with T1D and CD are seen more frequently in Arab countries compared to those with T1D only. Physicians of Group1 lean more towards frequent follow-up arrangement for people living with T1D and CD compared to physicians from Group2 (50% vs 21.5%, p-value 0.003). 54.5% of Group1 doctors think glycemic control is more difficult in children with both T1D and CD vs 21.5% of group2 who strongly agreed with that, p-value <0.001) and those with dual autoimmunity need more frequent follow-up by specialists (45% of group1 vs 22% of group2 who strongly agreed with that, p-value 0.02).

Both groups had similar views; forming multidisciplinary clinics to see patients would give better outcomes instead of seeing them separately by each specialist (t=0.740, p=0.460), starting a GFD in patients with T1D and concomitant CD reduces frequency of hypoglycemic episodes (t=0.863, p=0.389), adherence to GFD in people with CD and T1D is more challenging than in those with CD alone (t=1.452, p=0.148), no need for insulin dose increase in T1D and CD individuals (t=−1.861, p=0.064), the importance of having psychological support for them (t=−0.755, p=0.451) and the need for more detailed guidelines (t=0.338, p=0.735).

Conclusion: Glycemic control difficulty and follow-up needs are perceived differently between the two groups, physicians practicing in Arab countries would arrange more frequent follow up appointments. Both groups had similar views in terms of the effectiveness of GFD in reducing hypoglycemia, adherence challenges, and the need for psychological support and detailed guidelines.

ORAL ABSTRACTS

Evaluation of the Management of Diabetic Ketoacidosis in Children and Adolescents with Type 1 Diabetes at Two Hospitals In Mali

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Introduction: Diabetic ketoacidosis (DKA) is a metabolic state characterized by relative or absolute insulin deficiency and increased counterregulatory hormones that cause both hyperglycemia and ketone bodies production the accumulation of which leads to metabolic acidosis. It's a life-threatening emergency of type 1 diabetes (T1D) whose prevalence and incidence are increasing worldwide. Hypoglycemia, hypokalemia, and cerebral injury can occur during DKA management and risk can be reduced when pediatric treatment guidelines are followed. In Mali, DKA in children and adolescents is managed by adult endocrinologists who are not familiar whith guidelines specific to pediatrics. The objective of our study was to describe the management of DKA in children and adolescents with type 1 diabetes in Mali.

Methodology: This was a 7-month prospective and descriptive study conducted in two hospitals in Mali (Sominé DOLO hospital in Mopti and Mali hospital in Bamako) where there is neither a pediatric endocrinologist nor a pediatric intensive care unit. We included all children and adolescents up to 18 years of age with DKA defined as hyperglycemia > 200 mg/dl, serum bicarbonate < 18 mmol/l and ketonuria ≥ 2+ (ketonemia is not available in our setting). The severity of DKA was categorized by the degree of acidosis : mild (serum bicarbonate < 18 mmol/l), moderate (serum bicarbonate < 10 mmol/l) and severe (serum bicarbonate < 5 mmol/l). DKA resolution was defined by negative ketonuria.

Results: 24 patients were included. The mean age was 11.3 ± 4.9 years and the sex ratio was 0.5. 37.5% of the cases were debuting. Median glycemia was 3.85 g/L [IQR, 2.97-4.99]. DKA was mild and moderate in 70.8% and 29.2% of patients, respectively. No severe cases were observed. The median serum bicarbonate was 12 mmol/l [IQR, 9,07-13,42] and the median of anion gap was 18.6 mmol/l [IQR, 15,87-24,07]. The median of glycated hemoglobin was 14.5% [IQR, 12.2-16.9]. Insulin discontinuation in patients with established diabetes was the most common precipitating factor occurring in 60% of cases. The median amount of fluid, insulin and potassium administered was 474.3 ml/m²/h [IQR, 382.7-655.8], 0.3 IU/kg/h [IQR, 0.2-0.5], and 1.9 mmol/kg [IQR, 1.1-3.8] respectively. The median time to DKA resolution was 8 hours [IQR, 6-10]. Insulin was administered as an hourly 10 UI intravenous bolus because a continuous infusion device was not available. In addition, an initial 10 UI bolus was given intramuscularly. Hypoglycemia was the main complication observed during treatment, occurring in 20.8% of patients and hypokalemia occurred in one of four patients in whom serum potassium was monitored. We recorded one death due to cerebral injury in a newly diagnosed 9-year-old girl.

Conclusion: The management of DKA in children and adolescents in our setting is characterized by the administration of higher than recommended amounts of fluid and insulin, and unacceptably high rates of hypoglycemia, as well as 1 death in a population of 24 youth. An algorithm in line with recommendations and available local resources to prevent DKA management related complication is urgently needed.

Keywords: Type 1 diabetes, Children and adolescents, Diabetic ketoacidosis, Mali.

A Pediatric Clinical Case of Probable Sitosterolemia: a Challenging Diagnosis for a Rare Disease

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Introduction: Sitosterolemia is an autosomal recessive lipid disorder characterized by increased absorption and decreased biliary elimination of plant sterols or phytosterols, resulting in their accumulation in tissue and plasma. Markedly elevated phytosterol concentrations predispose to the development of premature atherosclerosis and early coronary artery disease. Therefore, an early diagnosis and prompt treatment are essential to prevent advanced atherosclerotic cardiovascular disease and to improve prognosis.

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A low sterol diet is the primary treatment for sitosterolemia, which differs from the recommendations for other types of dyslipidemias that may exacerbate this condition. We present the case of a pediatric patient diagnosed with probable sitosterolemia in a developing country, overcoming significant diagnostic and therapeutic challenges.

Case description: A 6-years 5-month-old male was referred to pediatric endocrinology with an 18-month history of painless tendinous lesions on his heels, associated with inflammatory erythematous papules on elbows and knees. Family history of lipid disorders was unknown. Physical examination showed tuberous xanthomas on his elbows and knees. Metabolic workup revealed severe hypercholesterolemia, with otherwise normal results. Gas chromatography could not be performed, therefore serum sitosterol levels were not measured. Initially, the patient was prescribed atorvastatin and ezetimibe. Whole exome sequencing (WES) revealed a pathogenic variant and a heterozygous variant of uncertain significance in the ABCG8 gene. The pathogenic variant (NM_022437.2:c.1083G>A) corresponds to a class 1 stop codon mutation, which leads to a change of tryptophan at position 361. The variant of uncertain significance (NM_022437.2:c.1598T>C), causes proline to be replaced by leucine in position 533. These results, along with clinical suspicion, led to the probable diagnosis of sitosterolemia. The statin was discontinued. Ezetimibe was continued along with a low-sterol plant diet. Consequently, the patient had a reduction in cholesterol levels (table 1) and a complete resolution of the xanthomas.

Discussion: Sitosterolemia is a rare autosomal recessive disease with wide phenotypic heterogeneity and unknown prevalence due to underdiagnosis (2). In our results, one of the alleles is pathogenic and the second allele is of uncertain significance (VUS), so we could not verify the diagnosis. Advanced methods such as gas chromatography could not be performed and genetic information from both parents was unavailable. Nevertheless, clinical suspicion and the patient's genetic results made the diagnosis of sitosterolemia probable. Proper treatment was then initiated, leading to clinical and biochemical improvement.

Conclusion: We report the case of a pediatric patient diagnosed with probable sitosterolemia in a developing country with some diagnostic challenges that were overcome. In fact, we are performing an in vitro expression functional assay, towards demonstrating pathogenicity potential and supporting the molecular basis for the clinical findings. The implementation of an appropriate treatment resulted in significant clinical and biochemical improvement. Finally, it is important to emphasize the need for access to genetic studies in our country.

Table 1. Patient's lipid profile evolution

Lipid profile (mg/dL)	2023			2024
	March	May	October	January
Total Cholesterol	729	473	199	226
HDL Cholesterol	43	38	51	51
LDL Cholesterol	660	420	134	161
Triglycerides	125	67	64	63

Genetic Testing Access and Results for Patients with Congenital Hyperinsulinism as Conducted through the CHI and University of Exeter Partnership

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Background and Aims: Congenital hyperinsulinism (HI) is the most frequent cause of severe, persistent hypoglycemia in newborn babies and children. Routine screening of the known etiological genes (n=>20) identifies a disease-causing variant in 40-50% of all cases. An accurate and timely genetic diagnosis is clinically important for all individuals as understanding the underlying genetic cause of the HI can guide the clinician in both medical and surgical management.

Congenital Hyperinsulinism International (CHI) is a nonprofit organization dedicated to improving the lives of those living with HI. Since 2018, CHI has provided funding to the University of Exeter to cover the cost of genetic testing for individuals who would otherwise be unable to receive genetic screening. This partnership not only aids individual families but also diversifies the pool of patients who are included in genetic databases which can aid in scientific discovery. This study aims to showcase the impact of enhanced genetic testing accessibility, emphasizing its contribution to a better understanding of genetic prevalence in HI, and to report on the results of the genetic testing conducted through this partnership.

Methods: Exeter recruited individuals with a clinical diagnosis of HI who were unable to access genetic testing through their own healthcare provider. Exeter performed rapid Sanger sequencing of the KATP channel genes, *ABCC8* and *KCNJ11*, in all individuals.

Targeted next-generation sequencing of the remaining known HI genes was performed in those without a disease-causing variant in the KATP channel genes if the HI persisted beyond 3 months or additional clinical features suggested syndromic-disease.

Results: Between July 2018 and December 2023 CHI funded genetic testing for 894 individuals from 61 countries with medically diagnosed HI. Routine screening identified disease-causing variants in 469 out of 894 individuals (52%). These variants were identified in 19 different genes. The most common genetic etiology was KATP channel HI, with *ABCC8* and *KCNJ11* variants identified in 360 patients (76.8% of solved cases). Of these, 238 individuals had bi-allelic or a dominantly acting variant confirming diffuse pancreatic disease. In 122 individuals, a paternally inherited variant predicted focal disease, which can be cured by lesionectomy.

Discussion: The partnership between CHI and Exeter has enabled 469 individuals with HI to receive an accurate genetic diagnosis. Understanding the underlying genetic cause of HI guides medical management by informing treatment decisions, prognosis, and recurrence risk within families. A global genetic access program helps ensure that individuals worldwide have access to crucial genetic testing, regardless of their socioeconomic status or geographic location. This access improves patient care by confirming genetic sub-types that aid in personalized treatment plans and contribute to the broader community by enhancing our understanding of genetic disease prevalence and providing a larger pool of data to identify new genetic variants of significance. By increasing access to genetic testing, this partnership is improving HI patient care around the world, providing novel insights into mechanisms of insulin secretion, and ensuring there are no barriers for children to receive testing regardless of where they are born.

An Adolescent Lives with Uncontrolled Type 1 Diabetes Mellitus Developed Acute Coronary Syndrome. A First Case Report from Oman in Pediatric Population

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Background: Type 1 Diabetes Mellitus (T1DM) is a prevalent chronic autoimmune disorder in pediatric populations, often leading to diabetic ketoacidosis (DKA) that necessitates emergency intervention.

Acute coronary syndrome with silent infarcts is a common and dreaded complication of type 2 diabetes; however, its incidence in long standing poorly controlled T1DM in children is relatively unknown. Patients with DKA typically present with symptoms of vomiting and abdominal pain, which are often managed with antiemetics like ondansetron or its analogues. Reports of coronary complications like vasospasm secondary to these drugs though rare may mimic ACS and should be recognised.

Case Presentation: We present a case of a 13-year-old female with a background of uncontrolled T1DM, attended the emergency department with a complaint of vomiting and hyperglycaemia, diagnosed to have mild DKA. She continued complaining of nausea and vomiting, for which she was given IV granisetron 2mg (A serotonin receptor (5HT-3 selective) antagonist). Soon after receiving the dose, she developed central chest pain radiating to the left side, and she felt lightheaded. Subsequent evaluation revealed significantly elevated troponin levels (359 ng/L, normal <14), and an ECG identified posterior STEMI features. Echocardiogram done a few hours later showed reduced ejection fraction of 35-40% with mitral and tricuspid valve regurgitation. The patient was managed in the intensive care unit, according to the ACS protocol, received aspirin, clopidogrel, and heparin. Few days later, the ejection fraction has improved and returned to normal with improvement of troponin level and normalising of the ECG. Patient was discharged home on her usual insulin doses with emphasis on the importance of controlling the diabetes to avoid the complication. A week later she was seen in the ambulatory follow up clinic, and she remained asymptomatic with good adherence to the plan. Her echocardiogram had normalised. To our knowledge, this is the first reported case of ACS in the context of uncontrolled T1DM in Oman among pediatric population.

Conclusion: For patients with uncontrolled T1DM, diligent follow-up and precise adjustment of medication are critical. It remains challenging managing diabetes among adolescents. Although ACS is rare in pediatric population, the severity of potential complications necessitates heightened vigilance for ACS in similar cases. This case raises the awareness of possible side effect of serotonin receptor antagonist and reinforces the need for further research to clarify the interactions between T1DM, DKA, and cardiovascular events. Early detection and appropriate management of ACS are essential for better prognosis and reducing both morbidity and mortality.

ORAL ABSTRACTS

Growth, Vitamin D and Calcium Profile in Diabetic Adolescent in Kinshasa. A Clinical Study

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Introduction: Adolescence is a period of life characterized by a rapid growth and development. Indeed, several factors participate in the growth phenomenon, including diet, hormonal action or the existence of chronic and metabolic pathologies. In literature, growth retardation, vitamin D and calcium deficiency are reported among adolescents living in areas where malnutrition is prevalent, particularly in sub-Saharan Africa. At the same time, growth abnormalities and trace element deficiency can be increased by type 1 diabetes mellitus. The objective of the study was to compare growth, the vitamin D and calcium profile in adolescents with type 1 diabetes melitus versus a control group in Kinshasa in order to assess the likely probable role of type 1 diabetes melittus on growth, vitamin D and calcium profile among adolescents in Kinshasa in context of malnutrition.

Methods: This was a prospective case-control study carried out over a 6-month period (February-July 2024) on a sample of 60 adolescents aged 13 to 17 years old. 30 adolescents with clinical type 1 diabetes mellitus were compared to a similar control group (no diabetic). Nutritional status was assessed by BMI for age, growth was assessed by the Tanner score, vitamin D « 25 (OH) D » and calcium were measured in serum. The normal value of vitamin D was between 30 and 70 ng/ml. The insufficiency was between 20 and 30 ng/ml, the deficiency for a value less than or equal to 20 ng/ml and the carence when < 10ng/mL. The normal calcium level was between 2.2 and 2.6 mmol/l, moderate hypocalcemia between 1.8 and 2.2 mmol/l and severe if the level is less than 1.8 mmol/l. The Mann-Whitney test was used to study associations, IC was 95%, p was significant if ≤0,05. Parathyroid hormone, growth factors and calcitonin were not measured because they were not available in the country.

Results: It appears from this study that the majority of diabetic adolescent (68.97%) had malnutrition compared to 44.83% in the control population [IC 95%] p=0.003. For the growth, 65.52% were stunted versus 31.03%, [IC 95%] p=0.001. Most diabetic adolescents (44.83%) had moderate hypocalcemia compared to controls (34.48%), [IC 95%] p=0.0003. In the diabetic group, vitamin D insufficiency was found in the majority of cases (42,17%) compared to 27.93% in the control population, and the deficiency in 22,5% versus 1,1% in control group, [IC 95%], p=0.0001.

The association between nutritional status and growth was significant, p=0.0003. The study also showed an association with growth, vitamin D and calcium status in adolescents with type 1 diabetes mellitus.

Conclusion: The study showed a significant negative impact of type 1 diabetes mellitus on growth, vitamin D and calcium status in diabetic adolescents in Kinshasa in context of malnutrition.

Limits of Study: No access to the dosage of parathyroid hormone, calcitonin and growth hormone because not available in the country and high cost.

POSTER ABSTRACTS

ADRENALS AND HPA AXIS

Establishing Colombia's First Community for Pediatric Congenital Adrenal Hyperplasia Patients

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Introduction: Congenital adrenal hyperplasia (CAH) is a rare autosomal recessive disorder caused by mutations in the CYP21A2 gene, leading to adrenal insufficiency, salt wasting, and virilization of the external genitalia. This condition imposes significant stress on patients and their families as they manage the disease and its extensive medical demands . In Colombia, factors such as national economic conditions, place of residence, education level of patients and caregivers, and health insurance coverage contribute to inequality in care. We recognized the need to establish a community where individuals living with CAH and their families could share experiences, build a support network, and receive education. A health needs assessment (HNA) was conducted to identify the community's most urgent needs and guide actionable steps.

Methods: A key alliance was formed between La Cardio Children's Hospital, CLAN*, the CAH Colombian Community, and the SiEndo Foundation**. With active community participation, a workshop was organized for patients and families affected by CAH. The agenda featured presentations by international and national experts and children had educational activities led by La Cardio Hospital's pedagogical teachers. A research protocol for a cross-sectional study was developed, including patient recall procedures, the meeting agenda, and study tools. Patient information was gathered on the RedCap platform, with approval from the La Cardio Ethics Committee (DDI 4917 - 2024).The team adapted the HNA from CLAN; a 112-question survey tailored for Colombia was finalized after pilot testing with six families. Attendees provided informed consent and completed the HNA for the research protocol: "Characterization and Assessment of Health Needs in a Congenital Adrenal Hyperplasia Pediatric Community in Colombia."

Results: The first Colombian CAH Patient Club meeting was held at LaCardio Hospital in Bogotá, with support from CLAN and SIEndo Foundation. Referrals from endocrinologists brought in 110 participants, including 32 patients and their families; 71.9% of patients were female and 28.1% were male, with the majority coming from urban residences. Although most families claimed their insurance covered treatment, they still noted medical accessibility was a big stressor for them. Other obstacles identified were lack of education about CAH, poor access to optimal health care and medications, and economic burdens. The data gathered from the HNA survey was transcribed to a RedCap database by La Cardio team and is under analysis. CLAN's CHECC (Child Health Equity Checklist Count) scorecard was applied; 7 families (21.8%) were identified to be at special risk and supportive actions implemented. Five months after the meeting, Colombia´s CAH community is functioning and growing with an active social media platform.

Conclusion: The club meeting led to the creation of the first Colombian CAH patient community. It became clear that CAH patients face enormous challenges. Bringing together the various stakeholders and beginning to address identified barriers allows participating providers to diversify their knowledge of their patient population and tailor their interventions for patients based on their unique circumstances. Future meetings should involve diverse backgrounds to improve perspectives and strategies.

Clinical and Genotypic Characterization of a Cohort of Male Patients with Adrenal Hyperplasia in a Pediatric Hospital in Cali

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Introduction: Congenital adrenal hyperplasia includes all hereditary disorders of cortisol adrenal steroidogenesis. Seven clinical forms of congenital adrenal hyperplasia are described according to the enzymatic deficiency; the most frequent is 21-hydroxylase deficiency. Classical form of 21 hydroxylase deficiency is characterized by severe impairment in cortisol and aldosterone synthesis (adrenal insufficiency) and hyperandrogenism (virilization of the affected female). Genetic diagnosis (CYP21A2) is essential and genetic counseling should be offered when necessary.. Suspecting it in time will prevent death from adrenal crisis. Treatment relies on the appropriate use of glucocorticoids and mineralocorticoids and multidisciplinary management.

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Objectives: To characterize male patients with adrenal hyperplasia assessed by pediatric endocrinology in a pediatric institution in Cali, Colombia.

Material and Methods: Descriptive observational study, retrospective case series type by reviewing medical records and data collection of male patients under 18 years of age with diagnosis of adrenal hyperplasia diagnosed between July 1, 2019 to June 30, 2021 in the Fundación Clínica Infantil Club Noel. RESULTS :Twenty-five men with diagnoses of adrenal hyperplasia (AHP) were included. A total of 14/25 patients (56.1%) had salt-losing ADH, these with adrenal crisis debut and with mean values at diagnosis of sodium 128.2± 11.0 mEq/L and potassium 6.4± 1.7 mEq/L, respectively. The diagnosis of HSP among salt losers was always made during the first 30 days of life, with a median age at diagnosis of 16.5 days (RIQ: 13.7 days to 30 days). In contrast, the age at diagnosis of nonlosers was older with a median of 3 years (RIQ: 3 months to 7 years). The median 17- hydroxyprogesterone (17-OHP) of 2000 ng/dL (RIQ: 1489 to 2000). 100% with macropenis and 88% with hyperpigmentation of genitalia and nipples and in (86.7%) body hypertrichosis. All salt-losing cases were treated with corticosteroids and fludrocortisone. For administrative reasons we were able to identify 52% of the associated genetic variants in CYP21A2 in 12 patients distributed as follows: three with c.844 G>T p.Val282Leu, three in Val 281 leu heterozygosis, four with Intron 2 , two with : c.518T>A, p.Ile173Asn heterozygous and one variant in CYP11B2 8q24.3 : c.594A>C ,p.Glu198Asp and in 10q22.1 : c.5237G>A p.Arg1746Gln in heterozygosis . All salt-losing cases were treated with corticosteroids and astonin.

Analysis: The importance of this study lies in the fact that there are no clear statistics in Colombia of adrenal hyperplasia and we must also be sensitized to its presence since it is potentially fatal in males since its diagnosis can be delayed by not having ambiguous genitalia, only macropenis. Therefore, it is important to suspect this clinical finding when we do not have neonatal screening for the disease and anticipate the onset of a crisis of severe saline loss, potentially lethal and avoid the associated morbidity and mortality

Adrenal Mass in a Pediatric Patient with Congenital Adrenal Hyperplasia: A Diagnostic and Therapeutic Challenge

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Introduction: Congenital adrenal hyperplasia (CAH) is a rare autosomal recessive disorder. The prevalence of adrenal masses in patients with CAH remains uncertain, and there is no established management guideline specifically for these lesions in pediatric populations. We present a case illustrating the diagnostic and therapeutic challenges encountered in a developing country when a patient with CAH develops an adrenal mass.

Case report: This patient is a 9 years and 11 months old female with 46 XX karyotype, diagnosed at the age of 3 months with classic salt wasting and genital virilization CAH, due to 3 variants in CYP21A2 gene (c.293-13 C>G heterozygous, p(?), probably pathogenic. c923dup, p.Leu308Phefs*6, heterozygous pathogenic. c1360C>T, p.Pro454Ser, heterozygous pathogenic). She had feminizing genitoplasty at one year of age. Despite being treated with high-dose hydrocortisone (24.8 mg/m2/day), fludrocortisone 0.1 mg/day, and good adherence, she remained poorly controlled. At the age of 7, she developed precocious puberty and is treated with triptorelin pamoate. At 9 years and 3 months, abdominal pain led to the discovery of a 22 mm right adrenal lesion on ultrasound. Four months later an abdominal CT scan revealed a 40 mm mass in the right adrenal gland with neoplastic appearance without calcification. A month later, the first MRIs done at a second level hospital reported an apparent decrease in the size of the mass. Subsequent MRI with adrenal protocol performed in a more experienced center, reported a lobulated image with regular edges, circumscribed, measuring 40x20x25 mm with heterogeneous enhancement in the late arterial phase that persists in the late equilibrium phase, without calcifications or necrosis. She had no features of Cushing's syndrome, and adequate androgen suppression was achieved with post-dexamethasone test; urine and plasma metanephrines were also normal. Right adrenalectomy was performed 8 months after the mass was diagnosed. Pathology confirmed a 4 cm adrenal cortical neoplasm compatible with adenoma without criteria of malignant behavior. Postoperative complications were absent, and abdominal pain resolved. Androgen control improved, allowing for a reduction in hydrocortisone.

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Discussion: Adrenal masses have been reported in up to 30% of patients with CAH. However, there is a paucity of literature on this association in children and adolescents. The most common adrenal lesions in CAH are myelolipomas, adenomas, hyperplasia, and, less frequently, carcinomas and pheochromocytomas. In this patient, the diagnostic process and imaging acquisition were delayed. The final MRI, performed with an appropriate protocol, did not definitively indicate a benign lesion due to its heterogeneous contrast uptake and prolonged washout time. Despite adequate family support and treatment adherence, the patient faced challenges in accessing close clinical follow-up and imaging at an experienced center. Given the progressive growth of the mass, surgical management was deemed necessary.

Conclusion: Adrenal masses in pediatric patients with CAH are diagnostically challenging. Clinical symptoms, proper imaging characterization of the lesion, and a multidisciplinary approach are key to appropriate management.

POSTER ABSTRACTS

DIABETES AND INSULIN

Characterizing Pediatric Type 1 Diabetes: Distinct Profiles of Cases with vs. without Complications in South Kalimantan, Indonesia

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Background: Diabetes mellitus type 1 (T1DM) in children represents a significant healthcare challenge, characterized by autoimmune destruction of pancreatic beta cells and necessitating lifelong insulin therapy. Despite advances in diabetes management, pediatric patients with T1DM remain at risk for a variety of complications, including ocular disorders, nephropathy and neuropathy. These complications can significantly impact the quality of life and long-term health of affected children. The variability in complication risk among children with T1DM poses challenges for clinicians in providing personalized care. Understanding the distinct clinical profiles of children with T1DM, particularly in regions like South Kalimantan, Indonesia, where healthcare resources and diabetes management practices may differ, is essential for optimizing treatment strategies and improving patient outcomes.

Aim: To identify and characterize the distinct profiles of pediatric Type 1 Diabetes with and without complication in South Kalimantan, Indonesia.

Methods: A retrospective studies was conducted using medical records of children aged >1 months old and <18 years old diagnosed with T1DM from January 2021-December 2023. Children who underwent eye examinations by ophthalmologists and were assessed for renal complications through albumin-creatinine urine ratio testing were included. Children who were newly diagnosed or had insufficient screening data were excluded from the study. Data collected from the medical records included baseline demographic information, laboratory results, and details of diabetes management and treatment. The data were then divided into two groups:those with complications (categorized as either ocular complications or nephropathy) and those without complications.

Results: A total of 37 patients were included in the study. The participants were divided into two groups based on the presence of complications. The group with complications 7 patients, who exhibited either ocular hypertension, juvenile cataracts, and albumin-creatinine greater than 30 mg/dL. The remaining 30 patients were classified as the group without complications. Children with complication groups had gender ratio of females to males 5:2 compared to 2:3 in without complication group. The mean age at diagnosis of this group was 6.7 (±2.4) years younger than 8.9 (±4.5) years in other group. The median duration of diabetes was 6.15 (3.3-9.2) years longer than without complication group 2.18 (1.1-2.4) years. The mean HbA1c level was 15.58% (±7.5%) notably high than other group 9.48% (±2.5%). The mean insulin dosage was 1.16 U/kg body weight per day in both groups.

Conclusion: Children with T1DM in South Kalimantan, Indonesia with complications exhibited a predominance of females, a younger age at diagnosis, a longer duration of diabetes, and higher HbA1c levels compared to those without complications. These findings underscore the critical need for effective glycemic control and emphasize the importance of frequent screening, even in asymptomatic individuals, to identify and manage potential complications early.

Epidemiological Evolution of Type 1 Diabetes in Children at the Mother and Child University Hospital Center of N'Djamena

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Introduction: Diabetes in children has undergone a profound change over the past twenty years worldwide. The change mainly affects children under the age of fifteen in the form of a real epidemic and at an increasingly early age of onset, posing serious problems in terms of care in pediatric settings. Diabetes imposes an economic burden on Africa, including catastrophic costs to combat the disease at the individual level. More than half of people with diabetes in the African Region are not diagnosed. Chad is so impacted because it has a high incidence of diabetes in children with increasing numbers.

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This study aims to present the epidemiological evolution of childhood diabetes through recruitment of new cases covered by the register of the pediatric department of the mother and child university hospital center of N'Djamena.

Materials and methods: The reference population is that of the Chari-Baguirmi department. The new cases of childhood diabetes came from public primary care units, private practitioners' offices, the CHU and peripheral hospitals in the Chari-Baguirmi department, at the origin of the recruitment of pediatrics quickly and systematically referred to the level of the pediatric department. All patients selected met the criteria for diabetes, as adopted by international organizations. The notion of insulin dependence was retained on weight loss, ketosis, and sometimes initial ketoacidosis and on permanent dependence on insulin treatment. Children with other forms of diabetes and rarer diabetes such as induced diabetes, Wolfram syndrome, Rogers's syndrome and monogenic diabetes were excluded from the analysis. The frequencies were established from the pediatric diabetes register of the pediatrics department. The recording was continuous with almost one hundred percent completeness. To calculate the incidence, all diabetic children meeting the diagnostic criteria and residing in the Chari-Baguirmi department for more than six months were included. The onset of diabetes considered is that of the first insulin injection. Incidence was expressed as the number of new cases per hundred thousand children. The incidence of those under fifteen years of age is expressed as an annual average of new cases over successive five-year periods, to mitigate the effect of annual fluctuations. To calculate the prevalence, all living children residing within the administrative limits of the department under the age of fifteen received in the pediatric consultation of the mother and child university hospital center were taken into account.

Results and Discussion: Over a period of four years, five hundred and forty-eight new cases of diabetes in children aged under fifteen meeting the diagnostic criteria were recorded in the department of Chari-Baguirmi with a considerable incidence rate. There is a gradual annual increase in the number of cases as noted by several authors. Conclusion The persistent increase in the incidence and age of onset of diabetes in this study is a reality. This development, mainly linked to the environment, poses new difficulties for families and teams responsible for the condition.

Assessment of Knowledge about Type 1 Diabetes in Children and Adolescents Living with Diabetes at the Nouakchott Mother and Child Hospital (Mauritania)

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Introduction: Patient education for the patient is a cornerstone and an indispensable tool in the management of people living with diabetes. Our objective was to assess the knowledge of type 1 diabetes among living patients with diabetes at the Mother-Child Hospital in Nouakchott, Mauritania.

Patients and methods: This was a cross-sectional, descriptive survey conducted from January 1 to March 31, 2024. All children and adolescents aged 10 years and older, living with diabetes who were seen or hospitalized during this period were invited to participate in the study. We used the knowledge assessment questionnaire developed by the 'Aide aux Jeunes Diabétiques' of France (1), which consists of 50 true/false questions of type True-False. We added the choice of 'I don't know'. The topics addressed were general about diabetes, self-monitoring, diet, hypoglycemia and hyperglycemia and management of insulin treatment. The level was considered low/insufficient when correct answers were less than 24/50, average if correct answers were between 25 and 39/50 and good or satisfactory when the number of correct answers is 40/50.

Results: A total of twenty-four patients were included in our study. The mean age was 14.35 ±3.45 years (10-22) . The mean age at discovery of diabetes was 10.97 ±3.61 years with a median of 10. The mean follow-up time was 3.37 years with a median of 2. The sex ratio was 1. The most represented age group was between 14 and 17 years old, with twelve patients or 50% and the least represented was over 18 years old with three patients (12.5%). The mean glycated hemoglobin was 10.43% (6-16.5%) with a median of 9.60%, a minimum of 6 and a maximum of 16.50. Diabetic retinopathy was diagnosed in only one patient, representing 4.1%. albuminuria was negative in all our patients. The average overall score for good answers was 27.92 (55.83%) with a minimum of 12 (24%) and a maximum of 37 (74%). None of our patients had a satisfactory or good level. However, 17 patients (70.83%) had an average level.

The level was insufficient in 7 (29.16%). The topics «Management of insulin treatment» and «Diet» were best understood by patients with 61.02% and 59.72% respectively of good answers. «General information about diabetes» and «Self-monitoring» had 56.94% and 50% correct answers, respectively. On the other hand, the topic «Management of hyperglycemia and hypoglycemia» was the least mastered by our patients with only a percentage of 46.52%.

Conclusion: The results of our study show that the overall level of knowledge about diabetes among our patients was average. These results should be a warning sign for an improve the level of knowledge of these patients. Further studies are needed to identify the weaknesses of therapeutic education to improve the management of young people living with diabetes in our country.

Keywords: Knowledge, Type 1 diabetes, therapeutic education, Nouakchott

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Eighteen Months of Effective Treatment with Golimumab Monotherapy in a Child with Newly Diagnosed Type 1 Diabetes Mellitus, Case Report and Review of Literature

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Type 1 diabetes mellitus (T1DM) is an autoimmune endocrine disorder. Despite new technologies in insulin administration and methods of measuring blood sugar levels, maintaining satisfactory levels of glycemic control is still a challenge. Thus, developing novel interventions to protect residual beta cell function, decrease insulin dependence, and enhance the general clinical profile of T1DM is particularly important.

The current case report describes a 5-year-old pediatric patient with newly diagnosed type 1 Diabetes Mellitus on 01/02/2023 who was treated successfully with Golimumab (TNF-α inhibitor) monotherapy without insulin for 18 months duration and remained near normoglycemic with reserved C-peptide level within the normal range. The previously asymptomatic patient presented with typical symptoms of polyuria and polydipsia and randomly obtained a blood sugar level of 315 mg/dL and an HbA1c of 6.5%. Anti GAD Abs (AntiGlutamic Acid Decarboxylase Antibodies) and Anti ZNT8 Abs (Anti-Zinc Transporter 8 Antibodies) were positive. The treatment rendered it possible to maintain near normoglycemic levels, preserve C-peptide, and defer the use of insulin without any side effects.

Low HbA1c with normal C-peptide levels were later noted following the start of golimumab therapy, and she remained near normoglycemic with time in range (TIR) within the target range of > 70% without insulin for 18 months. Thus, this example demonstrates that golimumab can improve beta cell function and maintain optimal glycemic control without insulin supplementation. In summary, during the 18-month treatment with Golimumab monotherapy without insulin, the C-peptide level was maintained within the normal range, the patient remained near normoglycemic, with TIR within the target of > 70% and HBA1c in the acceptable range of a maximum of 7.3%. Compared to other TNF-α inhibitors, including Infliximab and Etanercept, there are indications that these drugs may also protect beta cells. Still, the long-term advantages and risks may not be the same. Given the above findings, more comparative trials are still required to establish the optimum and least hazardous TNF-α inhibitor for T1DM patients. This case report suggests the possible treatment with a TNF-α inhibitor regimen, Golimumab, to manage newly identified T1DM sufferers. Based on such findings, the present study implies that early intervention with Golimumab can alter the disease progression by preserving beta cells from assault by the autoimmune system. Further research is needed to validate these effects and create a protocol of regular TNF-α inhibitors for T1DM treatment with the help of a large-scale clinical trial.

Table 1: Summary of HBA1c and C-peptide values from diagnosis throughout follow-up

Component	Ref Range	01/02/2023	09/02/2023	18/05/2023	22/08/2023	17/11/2023	10/3/2024	14/06/2024
HbA1C	<5.7 %	6.5	-	6.4	6.5	6.9	7.3	7.2
C-Peptide	0.37 - 1.47 nmol/L	0.48	0.48	0.99	0.55	0.73	-	0.65

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5 Months Infant with Recurrent Urinary Tract Infection Proved as Diabetic Ketosis without Acidosis: How to Diagnose and Treatment?

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Background: Neonatal Diabetes Mellitus (NDM) is a rare genetic disease with an incidence rate of 1:90,000 live births. NDM is often associated with severe complications, such as Diabetic Ketoacidosis (DKA), due to delayed diagnosis. Early diagnosis and treatment are essential to reduce these complications.

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Case: A 5-month-old baby boy was diagnosed with recurrent urinary tract infections. Over the past three months, his weight has decreased despite frequent drinking, and he often appears thirsty and urinates frequently.

On physical examination, the genital area, particularly the prepuce, appeared red, and upon applying pressure, smegma and pus were discharged from the external urethral orifice (OUE). Laboratory tests revealed leukocytosis (17,630 /uL), hyperglycemia (516 mg/dL), ketosis (1.6 mmol/L), and an HbA1c level suggestive of diabetes (11.0%). Blood gas analysis indicated compensated respiratory alkalosis. Based on the history, physical examination, and supporting tests, the child was diagnosed with ketosis in NDM accompanied by a urinary tract infection. A consultation with a pediatrician was conducted. The patient was given a blood sugar regulation protocol and administered fast-acting insulin at a dose of 0.1 units/kg bolus. Blood glucose levels were checked hourly, and ketones were measured every 2 hours until DM ketosis was resolved. The patient was monitored in the Intermediate Care (IMC) unit. The antibiotic Ampicillin-sulbactam was administered intravenously at a dose of 25-50 mg/kg every 6 hours. On the second day, the child appeared more active, and ketones were negative. On the sixth day, the child was discharged after the mother was instructed on how to use fast-acting insulin and administer oral sulfonylurea therapy at an initial dose of 0.3 mg/kg/day. Genetic testing was also planned for the patient.

Conclusion: Early diagnosis can be achieved through clinical manifestations accompanied by supportive laboratory tests. Expert consultation facilitates diagnosis, reducing the likelihood of complications.

Inaugural Diabetic Ketosis in a Child with Prader-Willi Syndrome

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Introduction: PRADER–WILLI syndrome is a genetic disorder caused by the absence of expression of certain genes present on chromosome 15.

Observation: This is an 11-year-old girl, originally from and living in Constantine, admitted for diabetic ketosis. On examination, the child presents with morbid obesity, with facial dysmorphism, and above all a significant growth retardation of -2DS

Discussion: Glycemic balance is obtained by an injection of basal insulin at night with boluses at meals after resolution of ketosis and given the frequency of hypoglycemia we switched to metformin , with very good glycemic balance, despite the use of GH.

Conclusion: Type 2 diabetes represents the major phenotype of diabetes in this syndrome; given all of these abnormalities, multidisciplinary management is necessary.

Complex Case of Diabetes Mellitus with Multiple Comorbidities

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Background: Diabetes mellitus is a chronic disorder which is common in children .Diabetes in children is usually Type 1 or monogenic or Type 2 diabetes mellitus. Diabetes rarely may be secondary to underlying insulin receptor defect or syndromic.

Case Presentation: M is 11 years old girl was born at term without any post natal complications .She has gross motor developmental delay , sensorineural hearing loss using hearing aids since age of 4 months of age , generalized seizure disorder on anticonvulsants since age of 8 month and early pigmentary retinal changes and alternating esotropia. She also has Type IV renal tubular acidosis with hypertension on atenolol and hydrochlorothiazide. She was admitted at 3 years of age with polyuria and hyperglycemia without any ketoacidosis .She was thought to have either Monogenic diabetes or Diabetes secondary to mitochondrial cytopathy. She was started on MDI insulin regime ,Glargine and Actapid. Her Diabetic antibodies resulted positive for Anti GAD but C-peptide level was 1.80 ng/mL .Her genetic testing showed Pathogenic mutations in the PRKDC gene cause immunodeficiency type 26, with or without neurologic abnormalities (OMIM #615966). Her Anti TPO > 500 IU/l came positive during routine yearly evaluation of other autoimmune diseases. However, she remain clinically & biochemically euthyroid .she presented recently at 11 years of age with goiter and irritability ,her TFT showed high fT4 and Suppressed TSH and so she is started on Carbimazole .Her Diabetes is controlled with HB A 1 of 6.1 on Insulin 1.2 U/kg/day .She has a sister with similar genetic mutation and phenotype but with out Diabetes mellitus or Hyperthyroidism.

Conclusion : This patient with multiple endocrine and non endocrine problems .Neither the clinical phenotype nor PRKDC gene mutation explain her disease. Diabetes mellitus & Hyperthyroidism are due to autoimmunity , however etiology of Type 4 RTA ,Global developmental delay ,SNHL and seizure disorder can not be explained by one disease process .Open Question to all ; Are we missing any new genetic mutation or something else?

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DISORDERS OF SEXUAL DEVELOPMENT

The Importance of Detailed Clinical Characterisation in Reaching a Definitive Diagnosis after Next-Generation Sequencing (NGS) Genetic Testing in Patients with 46,XY DSD

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Introduction: DSD is a genetically heterogeneous group of disorders. NGS has transformed the diagnostic landscape, enabling simultaneous analysis of multiple genes. Nevertheless, interpreting NGS results can be challenging due to the lack of DSD-specific guidelines for variant classification. Deep phenotyping, involving a detailed clinical characterisation, is crucial in optimising variant interpretation and aetiologic diagnosis. Specifically, the application of phenotype-related PS4, PP4, PS2/PM6 and PM3 labels from the ACMG guidelines are crucial to classify variants as likely pathogenic (LP) or pathogenic (P). However, they are infrequently applied when using online interpretation tools (e.g. Franklin, Varsome), which may lead causative variants to be classified as Variants of Uncertain Significance (VUS).

Aim: This study aims to evaluate the impact of deep phenotyping on variant interpretation and aetiological diagnosis in patients with 46,XY DSD. We will specifically assess the role of phenotypic specificity in applying the PS4, PP4, PS2/PM6 and PM3 labels, improving the classification of variants and reducing the number of VUS.

Methods: A cross-sectional study was conducted including patients with 46,XY DSD. A comprehensive clinical evaluation was performed, including physical examination, hormonal laboratory and imaging studies. Subsequently, genetic testing by NGS was performed and cases in whom candidate gene variants were identified were included. Our institutional Clinical Genomics team classified the candidate variants following ACMG guidelines and ClinGen Working Group recommendations, with a particular focus on the phenotype-related labels PS4, PP4, PS2/PM6 and PM3. The resulting classifications were compared with those returned by the online interpretation tools Franklin and Varsome.

Results: 13 patients with 46,XY DSD were included. Clinical evaluation allowed for the classification of the cases into the following presumptive diagnoses: gonadal dysgenesis (n=7), and defects in androgen synthesis (n=3) or action (n=3). NGS results identified 15 variants in the 13 patients. In the 7 cases with gonadal dysgenesis, we identified 1 variant each in DHX37, MYRF and WT1 genes, 2 variants in NR5A1, and two copy number variants (CNVs): 1 in chromosome 3 and 1 in chromosome 9. In the 3 cases with androgen synthesis defects, we found 2 variants in HSD17B3 and in SRD5A2, and 1 in POR. Finally, in the 3 cases with androgen action defects, we detected 3 variants in AR. Following ACMG guidelines and ClinGen recommendations, with a particular focus on the phenotype-related labels PS4, PP4, PS2/PM6 and PM3, 11 variants were classified as Pathogenic, and 4 as Likely Pathogenic. When loaded into the online tools Franklin and Varsome, the same 15 variants were classified as follows: Pathogenic (Franklin: 7; Varsome: 3), Likely Pathogenic (Franklin: 5; Varsome: 8), VUS (Franklin: 3; Varsome: 3), and 1 with no returned result (Varsome: 1), as detailed in Table 1. As compared to Franklin, we reclassified 3 VUS variants as LP or P, and 3 LP as P. As compared to Varsome, we reclassified 3 VUS as LP or P, 6 LP as P, and 1 without results to P.

Conclusion: The exhaustive clinical evaluation improved the aetiological diagnostic yield of NGS results in patients with 46,XY DSD.

Table 1. Description of Variants Identified in 13 Patients with 46,XY DSD and their Classification by the Clinical Genomics Team, Franklin, and Varsome.

Case N°	46,XY DSD Presumptive Diagnosis	Variant description	Classification by Institutional Clinical Genomics team	Classification by Franklin	Classification by Varsome
1	Gonadal dysgenesis	***NR5A1*** Heterozygous NM_004959.5:c.259C>T NP_004950.2:p.(Arg87Cys)	LP	LP	LP
2	Gonadal dysgenesis	***NR5A1*** Heterozygous NM_004959.5:c.52del; NP_004950.2:p.(Asp18ThrfsTer57)	P	LP	LP
3	Gonadal dysgenesis	***DHX37*** Heterozygous NM_032656.4:c.2021G>A NP_116045.2:p.(Arg674Gln)	LP	LP	VUS
4	Gonadal dysgenesis	***WT1*** Heterozygous NM_024426.6:c.643C>T p.(Gln215*)	P	LP	LP
5	Gonadal dysgenesis	***MYRF*** Heterozygous NM_001127392.3:c.965G>A NP_001120864.1:p.(Trp322*)	P	P	LP
6	Gonadal dysgenesis	Heterozygous **CNV** prediction of 2.7 Mb on chromosome 3	P	P	P
7	Gonadal dysgenesis	Heterozygous **CNV** prediction on chromosome 9	P	P	No result
8	Defects in androgen synthesis	***POR*** Homozygous NM_000941.3:c.262G>A NP_000932.3:p(Gly88Ser)	P	VUS	VUS
9	Defects in androgen synthesis	***HSD17B3*** Compound heterozygous 1°) NM_000197.2:c.278-1G>C	P	P	P
		HSD17B3 Compound heterozygous 2°) NM_000197.2:c.377G>A NP_000188.1:p.(Gly126Glu)	LP	VUS	VUS
10	Defects in androgen synthesis	***SRD5A2*** Compound heterozygous 1°) NM_000348.4:c.680G>A NP_000339.2:p.(Arg227Gln)	P	P	LP
		SRD5A2 Compound heterozygous 2°) NM_000348.4:c.721T>C NP_000339.2:p.(Tyr241His)	LP	VUS	LP
11	Defects in androgen action	***AR*** Hemizygous NM_000044.6:c.1078C>T NP_000035.2:p.(Gln360*)	P	LP	LP
12	Defects in androgen action	***AR*** Hemizygous NM_000044.6:c.2376_2377dup NP_000035.2:p.(Gln793LeufsTer17)	P	P	LP
13	Defects in androgen action	***AR*** Hemizygous NM_000044.6:c.1768+1G>A	P	P	P

POSTER ABSTRACTS

FAT, METABOLISM, AND OBESITY

Pediatric dyslipidemia; experience from resource limited settings in Pakistan

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Introduction: The abstract aims to highlight the spectrum of pediatric lipid disorders as seen in the first lipid clinic for children.

Method: the data was collected over two years 2022-2024 from the pediatric lipid clinic for children up to 16 years of age.

Results: A total of 29 children were seen with different dyslipidemias. Age ranges from neonate (19days old) to 13 years. Females were 15 and males were 14. Familial hypercholesterolemia was the most common disorder seen in 21 patients. Consanguinity was seen in 80% of the cases. Out of these 14 (48%) had homozygous familial hypercholesterolemia (HOFH). 7 (24%) were heterozygous familial hypercholesterolemia. Mean cholesterol levels were 512mg/dl. HOFH patients were mainly diagnosed on the basis of tendon xanthomas and lipemic samples as seen in figure 1.

7(24%) had primary hypertriglyceridemia including two with familial hyperchylomicronemia syndrome. Pancreatitis was reported in 4 patients. One patient had abetalipoprotienemia. Non-availability of genetic testing and treatment options other than statins and ezetimibe are the major burden in the management of pediatric dyslipidemias.

Conclusion: pediatric dyslipidemias remain underdiagnosed worldwide more so in resource constraint settings.

Table 1: lipoprotein electrophoresis analysis used in primary hypertriglyceridemia

Patient	Alpha (23-53%)	Pre- beta (5-22)	Beta (39-70)	Chylomicron (0-2%)	type
1	23.4	41.2	32.3	3.1	Type V (hyper pre beta+chylomicrnemia)
2	30.2	17.8	41.8	10.2	Type 1 (hyperchylomicronemia)
3	8.9	5.4	84.5	1.2	Type II A (hyper betalipoproteinemia
4	26.2	25.9	47.3	0.6	Type IV (hyper pre beta lipoproteinemia)
5	8.9	42.5	45.5	3.1	Type V
6	12.8	49.9	32.7	4.6	Type V
7	34	18	37.5	10.5	Type 1

Table 2:

Lipid Profile of patients with primary hypertriglyceridemia

patient	age	gender	Triglyceride	cholesterol	LDL-C	HDL
Patient 1	male	4 years	1750	73	16	9
Patient 2	male	1 years	1600	93	5	8
Patient 3	female	10 months	1428	166	135	12
Patient 4	female	4.7 year	240	148	78	35
Patient 5	male	4.3 year	4235	131	5	10
Patient 6	female	7 years	3103	229	9	15
Patient 7	male	4 years	1750	158	9	6

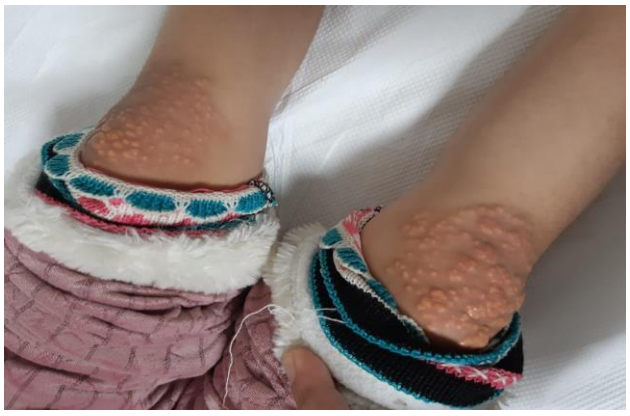

Figure 1.

Optimising the Clinical Assessment of Childhood and Adolescent Obesity in Jordan

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Background: Clinical practice guidelines aim to facilitate early diagnosis, implement early treatment and minimise gaps in care regardless of physician expertise or level of seniority. Consensus guidelines must be adapted to meet local and regional differences allowing for optimum benefit with what is available in terms of diagnostics and treatments. Childhood and adolescent obesity are no exception to this.

Aims: In this survey we aim to understand practice variation and gaps to better advocate for a regional and local care plan for obesity care in the young. *Methods:* A questionnaire was conducted between July 2024 and August 2024, aimed at healthcare professionals of all medical specialties. It consisted of 104 questions.

Results: Family medicine consultants treat most obese youth in Jordan. Several gaps were identified in the assessment and care of patients, including but not limited to psychosocial effects of obesity and efficient MDT work.

Conclusion: clear consensus guidelines are necessary to tackle childhood and adolescent obesity in Jordan.

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FETAL, NEONATAL ENDOCRINOLOGY, AND METABOLISM

Transforming Neonatal Care in Pakistan: A Clinical Audit of Congenital Adrenal Hyperplasia Screening

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Objective: This study aims to evaluate the effectiveness and efficiency of weight-based threshold levels for 17-hydroxyprogesterone (17-OHP) in screening newborns for 21 hydroxylases deficiency-congenital adrenal hyperplasia (CAH)

Design: Since January 2021 CAH screening was incorporated into the ongoing newborn screening program at Aga Khan University Hospital Karachi Pakistan,17OHp was assayed through Spectro fluorometry of Dried blood spots obtained via heel prick. The cutoff levels of 17OHP were determined based on the birth weight and age of the sampling.

Results: From January 2021 to July 2024, 20992 newborns were screened for CAH out of a total of 23533 births, only one CAH patient was detected, and therapy was started at a median age of 7 days. 2 patients were detected due to family history of sibs for CAH were not included in this cohort. Over the years we have seen a decline in the false positive rate from 1.2% to 0.06% to date, no false-negative cases have been detected.

Conclusion: Employing weight-based standards to assess 17-OHP levels during the screening process for 21-OH-D-CAH has been found to significantly lower the number of false-positive outcomes**.** The effectiveness and efficiency of utilizing this method for screening newborns for 21-OH-D-CAH will be determined through extended follow-up of the screened population.

Abnormalities of Sexual Development at CHU Sylvanus Olympio: Epidemiological, Diagnostic and Medical Aspects

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Introduction: Anomalies of Sexual Development designated by the acronym DSD (Disorders of Sex Development) in Anglo-Saxon literature constitute a large group of congenital anomalies of the urogenital tract and reproductive system affecting sexual determination and/or differentiation.

The main objective was to describe DSD, determine their classification, determine the aetiology and describe therapeutic strategies in a hospital environment in Lomé.

Method: this is a retrospective study on a series of cases of DSD seen in endocrinology consultation at Sylvanus Olympio University Hospital over a period of 36 months, from January 1, 2020 to December 31, 2022.

Results: A total of 6 patients met our inclusion criteria. The average age was 5.33 years with extremes of 1 and 14 years. Five of the 6 patients were raised as girls. The main reason for consultation in our study was the atypical aspect of external genitalia. Female type karyotype was found in 4 patients and male type in 2. In 46 XX DSD patients, congenital adrenal hyperplasia due to 21 hydroxylase block and ovotestis were the aetiologies found while 3βhydroxysteroid dehydrogenase block and partial androgen insensitivity were the aetiologies found in 46 XY DSD patients. None of the patients had performed the molecular genetic test for diagnostic confirmation. Medical treatment with hydrocortisone was performed in 3 patients and 2 patients benefited from androgen treatment.

Conclusion: this study made it possible to note the delay in the diagnosis of DSD, the interest in medical treatment and the need for a multidisciplinary approach in the management of DSD cases.

Keywords: DSD, karyotype, hospital environment, Lomé

Golden Phase for Optimal Treatment. Recombinant Gonadotropin Therapy During Mini puberty in Males with Hypogonadotropic Hypogonadism: A Case Series

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Introduction: The hypothalamic-pituitary-testicular (HPT) axis is highly active in healthy male newborns until 3-6 months of age. This phase, known as ‘minipuberty,’ is characterised by follicle-stimulating hormone (FSH) stimulation of Sertoli cells, leading to increased testicular volume and elevated serum levels of anti-Müllerian hormone (AMH) and inhibin B.

Additionally, luteinising hormone (LH) promotes testosterone production in Leydig cells, contributing to normal penile growth and testicular descent into the scrotum. In congenital hypogonadotropic hypogonadism (CHH), minipuberty is absent due to a deficiency in gonadotropins. While the standard treatment involves testosterone replacement, recent literature on the use of recombinant gonadotropins to simulate minipuberty is promising. This study evaluates the effects of recombinant FSH (r-FSH) in combination with human chorionic gonadotropin (hCG) or recombinant LH (r-LH) in male infants with CHH or combined pituitary hormone deficiency (CPHD) treated during minipuberty.

Methods Study Design and Location: A longitudinal, retrospective study of a UK cohort of male infants with CHH, treated during minipuberty at Barts Health NHS Trust. Objective: To assess changes in penile size, testicular volume and position, and serum levels of LH, FSH, testosterone, AMH, and inhibin B, as recorded in a bespoke REDCap database.

Results: Five male infants with CHH or CPHD received recombinant gonadotropic therapy during minipuberty (median age 0.6 years; range 0.5-0.9 years). All patients presented with micro-penis and micro-orchidism (testicular volume ≤0.4 mL as measured by ultrasonography), and four had cryptorchidism. No adverse effects were observed. Following treatment with r-FSH combined with hCG or r-LH, significant increases were observed in penile length (median increase 12.5 mm; range 2.0-25 mm), testicular volume (median increase 0.3 cc; range 0.1-1.1 cc), serum FSH (median increase 7.6 IU/L; range 3.6-55.0 IU/L), LH (median increase 4.5 IU/L; range 0.7-5.8 IU/L), testosterone (median increase 4.5 nmol/L; range 0.6-9.2 nmol/L), and inhibin B (median increase 256 pg/mL; range 36.9-605.1 pg/mL). Post-therapy improvements in testicular position were more variable, with two patients showing descent into either the scrotum or distal inguinal canal.

Discussion: Our findings suggest that recombinant gonadotropic therapy during minipuberty has the potential to provide significant clinical benefits for infants with CHH, by stimulating Sertoli cell population and resolving micro-penis. In this small series, the best outcomes were observed with a four-month regimen of combined r-LH/r-FSH therapy. The use of combined gonadotropins may enhance future fertility potential in infants with CHH; however, long-term follow-up data are needed.

POSTER ABSTRACTS

GROWTH, GH and IGFs

Understanding Stunted in School-aged Children During Covid-19 Pandemic: Cross-Sectional Study from Indonesia

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Objectives: Linear growth failure in childhood is the most prevalent form of undernutrition globally. Over 165 million children under five are affected by chronic malnutrition or stunting. Despite a declining prevalence, approximately 30,8% or 27.000 children under five and 23,6% or 40.000 school-aged children remain stunted. Beyond impaired physical growth, stunting has multiple pathological changes marked by linear growth retardation in early life, associated with increased morbidity and mortality, reduced physical, neurodevelopmental and economic capacity and an elevated risk of metabolic disease into adulthood. The intergenerational cycle continues as stunted women often have stunted children, perpetuating poverty and reduced human capital that is difficult to break, especially during Covid-19 Pandemic. This study aims to determine the prevalence and risk factor of stunted among Indonesian school-aged children and its relationship with socio-demographic variables during Covid-19 Pandemic to develop strategic interventions.

Methods: A cross-sectional study with a consecutive sampling of 449 children aged 6-8 years old attending primary schools in Cirebon City, West Java, Indonesia from April – November 2021. We asked caregivers to perform measurements using a measuring tape in Frankfort plane based on the researchers' video instructions. We performed bivariate analysis using Chi-Square or Fisher's exact test to determine the socio-demographic variables associated with stunted.

Results: The prevalence of stunted was 23.8% (n=107). The analysis of caregiver characteristics revealed that most were young adults (31,3%, p=0,240) with low educational levels, particularly fathers (35,3%, p=0,006) and mothers (33,0%, p=0,019). Additionally, many fathers (30,3%, p=0,487) and mothers (24,2%, p=0,851) were unemployed, and a significant proportion had low socioeconomic status (35.5%, p=0.000). Subject characteristics showed that most were female (25,8%, p=0,396), aged 6 years (20,2%, p=0,206), with a history of premature birth (25,9%, p=0,976), low birth weight (27,6%, p=0,791), and normal delivery (26,1%, p=0,085). A notable finding was the history of exclusive breastfeeding (29,9%, p=0,006) for more than two years (33,1%, p=0,001) and delayed introduction of complementary feeding beyond one year of age (40,0%, p=0,025).

Conclusions: A significant proportion of Indonesian elementary school children were stunted, particularly during the COVID-19 Pandemic. Growth is largely regulated by hormonal signals that are highly responsive to nutritional status, requiring both macro- and micronutrients for normal development. Nutritional deficiencies impair growth, and stunted is strongly linked to sociodemographic factors such as caregiver education, income and challenges in providing adequate nutrition. Addressing stunted requires targeted interventions, including education and training for parents and caregivers on proper nutrition, particularly in exclusive breastfeeding and introduction of complementary feeding, while incorporating these sociodemographic factors into strategic solutions.

Keywords: Stunted, Stunting, Prevalence, Risk factor, COVID-19 Pandemic.

The Relationship between Parents' Education and Economic Levels and Growth in Children with Thalassemia

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Background: The coordinated action of several intricate regulatory systems in different ways leads to child growth. Every child has a genetic root and a certain capacity for growth, which can be influenced by various variables during prenatal and the postnatal period. The influence of SEPE (Social-Economic-Political-Emotional) factors on growth has been extensively studied in the general population of healthy children. But on the other hand, in children with chronic diseases, the underlying condition is still believed to be the cause of growth disorders.

Objectives: The aim of this study is to analyze the correlation between parents' education and economic levels and growth in children with transfusion dependent thalassemia.

Methods: A total of 61 children with transfusion dependent thalassemia were included. Children and parents completed a questionnaire, and data on weight, height, and BMI were collected. Ferritin data was obtained from the most recent laboratory examination in the medical record. Anthropometric analysis using WHO growth chart age 5-19 years old using Z score standard deviations (SDS). Statistical analysis used were St. Nicholas House Analysis (SNHA) for a first impression of the data. Spearman correlation test and a multivariate regression model. For data analysis, we used RStudio, an open-source program with $p < 0.05$ being considered significant.

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Results: The average of height SDS is -2.281 ± 1.42, duration of illness is 95.17 ± 49.4 months, and serum ferritin level is 2709.41 ± 2221 ng/mL. The correlation test and multiple linear regression analysis showed no correlation of father's education, mother's education, and salary with height SDS with (rho=0.07; p=0.59), (rho=0.1; p=0.44), and (rho=0.16; p=0.19), respectively.

Conclusions: There is no correlation between parents' education and salary with height SDS in children with transfusion dependent thalassemia. Further research is needed to explore the relationship between SEPE factor and growth in children with underlying chronic disease.

POSTER ABSTRACTS

MEDICAL EDUCATION

Building Global Citizenship with Human Rights for Minors in Transition to Adulthood and Its Impact on the CITIZENSHIP STATUTE with the GEFENANOCAPADOFILIA METHOD and HISTOLOGY ALGORITHM REGOCI

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Introduction: To raise awareness about medical responsibility and its administrative and political aspects in age-focused citizenships and their transitions in migrant phenomena, evaluating the impact of the CITIZENSHIP STATUTE, the GEFENANOCAPADOFILIA METHOD, and the HISTOLOGY ALGORITHM REGOCI.

Methods:
Approach in the medical history
Criteria for determining the accurate sex of a newborn
Histology clinical algorithm
Narrative of gender inconformity/incongruence related to sexual health in children and adolescents

Results:
Pre-existence of publications on emblematic cases
Established protocol in jurisprudential rule with REGOCI

Conclusions: To raise awareness among international human rights organizations about the CITIZENSHIP STATUTE, GEFENANOCAPADOFILIA METHOD, and HISTOLOGY ALGORITHM REGOCI. Glossary (Lexicography of Expression Philology) CITIZENSHIP STATUTE: A concept that serves to raise awareness in matters of genotype, phenotype, civil status <>, nationality, name, capacity, assets, domicile, and affiliation. GEFENANOCAPADOFILIA: A thought framework that encompasses and raises awareness in the words genotype, phenotype, civil status <>, nationality, name, capacity, assets, domicile, and affiliation in the construction of global citizenship with human rights for minors transitioning to adulthood and its impact on the CITIZENSHIP STATUTE. HISTOLOGY ALGORITHM REGOCI: A method and analysis of civil gonadal recognition in newborns before the annotation of the sex component in the citizen file or sensitive data assignment in the identification of the biological marker.

POSTER ABSTRACTS

MULTISYSTEM ENDOCRINE DISORDERS

Sociodemographics, Clinical-Endocrinological and Psychological Characterization of Series of Pediatric Cystic Fibrosis Patients in Colombia

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Introduction: Cystic fibrosis (CF), caused by mutations in the CFTR gene (7q31.2), affects an estimated 1 in 2,500-3,500 individuals globally (1,2). While primarily impacting the respiratory and gastrointestinal systems, treatment advances have extended life expectancy but increased multisystem complications. Recent research emphasizes endocrinological manifestations (EM) in CF, including CF-related diabetes (CFRD), growth retardation, and low bone density, which significantly affect quality of life and prognosis (3). Understanding these complications is crucial for optimizing treatment, necessitating personalized care from a multidisciplinary team at specialized CF centers. In Colombia, detailed characterization of CF patients is limited. This study aims to describe the sociodemographic, clinical-endocrinological and psychological features of pediatric CF patients.

Methods: This case-series study included CF patients treated by pediatric endocrinology at LaCardio's CF care-center in Bogotá from 2015-2022. Ethics committee approval (CEIC-055-2024) was previously obtained. Data from health records were collected via the REDCap platform and analyzed descriptively, focusing on sociodemographic, primary-secondary diagnosis, anthropometric, and clinical variables, particularly EM.

Results: Patients and Demographics: A total of 28 patients (50% female) were included. The median age at diagnosis was 24 months (IQR 12-96), with 64.3% (n=18) diagnosed after 12 months. The F508 mutation was present in 25% (n=7) of patients. Socioeconomically, 89.3% (n=25) lived in urban areas, and 67.8% (n=19) were in lower-middle-to-lower socioeconomic stratum.
The median distance to the care-center was 22.0 km (IQR 31.5-96). Family types included single-parent (n=9) and nuclear (n=8). Endocrinological Manifestations: At initial consultation, 57.1% (n=16) of patients had EM, with hypovitaminosis D being the most common. See Table 1. By the latest consultation, 82.1% (n=21) had at least one EM.

Mental Health and Developmental Issues: Of the 28 patients, 39.2% (n=11) had a mental health diagnosis, with 63.6% (n=7) of them being female. Anxiety was most common (25%, n=7), followed by depression (17.9%, n=5). While 10.7% (n=3) were medicated. Developmental delay was observed in 14.3%, and 39.2% faced academic difficulties, particularly among females (58.4%).

Table 1: Frequencies of EM in Initial Evaluation

EM	Frequency (%)
Hypovitaminosis D	15 (53%)
Short Stature	2 (7.1%)
Dyslipidemia	7 (25%)
Puberal Disorder	2 (7.1%)
Hypothyroidism	2 (7.1%)
CFRD	2 (7.1%)
Osteopenia	3 (10.7%)

Discussion: This study describes EM in a series of Colombian pediatric CF patients, showing a higher prevalence of 82.1% in contrast to 30-40% globally (4), The primary EM identified were hypovitaminosis D, short stature, and dyslipidemia. However, the descriptive nature of the study and the limitations associated with case series restrict the broader applicability and interpretability of these findings. Further research in Colombia should focus on endocrinologic manifestations, genetic profiles, mental health and sociodemographic implications to enhance public health initiatives and care for CF patients.

Association Between Congenital Adrenal Hyperplasia and Diabetes in Children

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Background: There were few reported cases of association of diabetes with congenital adrenal hyperplasia (CAH). CAH due to 21 hydroxylase is due to biallelic mutation at CYP21A2 gene at chromosome 6.p21.33, lies in close proximity to the HLA B and HLA DR loci, at chromosome 6p21.3 which are implicated in type 1 diabetes (T1D). On the other hand, it is known that patients who are on glucocorticoid therapy are at risk for metabolic syndrome, with alteration of glucose metabolism and increased level of insulin resistance.

Methods: A 9-year-old female patient who was diagnosed with CAH during neonatal period and who was on hydrocortisone and fludrocortisone, presented to a peripheral hospital with fever, vomiting and abdominal pain and was managed there as a case of adrenal crisis. Later she was transferred to our hospital. She had decreased level of consciousness and was severely dehydrated.

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Results: Her blood glucose was 400 mg/dL, and metabolic acidosis. She was given normal saline bolus and stress dose hydrocortisone. Corrected blood sodium 123 mEq/L, and potassium 5.9 mEq/L. She was started on intravenous hydration, but she kept having hyperglycemia and metabolic acidosis. Urine ketones was +3. Then she was started on intravenous insulin infusion in addition to the intravenous fluid as management for diabetic ketoacidosis (DKA). There was no significant history of polyuria or polydipsia, but she was noticed to have polyuria during the initial period of admission. DKA resolved within less than 24 hours. HbA1c was 12% ,

Conclusions: The association between CAH and diabetes is under-reported in pediatric population. Physicians should have high index of suspicion of diabetes when a child with CAH presented with decreased level of consciousness, metabolic acidosis and polyuria, and not solely focus on acute management on adrenal crisis management.

POSTER ABSTRACTS

PITUITARY NEUROENDOCRINOLOGY, AND PUBERTY

Pituitary Stalk Interruption Syndrome Presenting in a 4 years Old Girl with Recurrent Hypoglycaemia and Developmental Delay: A Case Report

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Introduction: Pituitary stalk interruption syndrome (PSIS) is a rare congenital pituitary anomaly. Classical neuroimaging findings are thin or interrupted stalk, absent or hypoplastic anterior pituitary gland, and/ or an ectopic posterior pituitary. The diagnosis can be delayed if the signs and symptoms of PSIS during childhood are often overlooked. The permanent anterior pituitary hormone deficiencies appear gradually during childhood and can progress into panhypopituitarism during adulthood.

Method: Here, we report a case of a four years old girl, who first presented with symptomatic hypoglycaemia at the age of 2 years old.

Case: She had a generalized seizure with random blood sugar of 2.9 mmol/L at 2 years old, which was managed with intravenous dextrose infusion. She was not syndromic and had no midline defect. The family defaulted on the subsequent clinic appointment. She had intermittent hypoglycemia at home, whereby her parents treated with sugary drinks. She presented again at 4 years old with another episode of symptomatic hypoglycaemia. She was markedly short (height SDS -5,26 SDS) with cherubic facies and global developmental delay. Extensive endocrine workups showed low serum growth hormone, inadequate adrenal response during the ACTH stimulation test and central hypothyroidism. MRI findings were consistent with PSIS. Introduction of growth hormone therapy alleviated her hypoglycaemia. Together with thyroxine and hydrocortisone replacement, she demonstrated catch-up growth and improvement in her developmental milestones.

Critical sampling	Results	Reference range
RBS	2.6 mmol/L	
Blood ketone	0.5 mmol/L	
Blood gas	pH 7.47, pCO2 29 mmHg, HCO3- 21.1 mmol/L, BE -2 mmol/L	
Insulin	6.25 pmol/L	17.8 - 173
C-peptide	129 pmol/L	366.66 - 1466.65
Growth hormone	0.062 mcg/L	0.050 - 5.11
Cortisol	270 nmol/L	
Lactate	1.04 mmol/L	
Ammonia	46 mmol/L	

Conclusion: Prompt recognition of hypoglycaemia as the marker of pituitary hormone deficiency and referral to Paediatric Endocrinology service are vital. Early hormonal intervention will lead to better outcomes.

Fahr's Syndrome as a Manifestation of Autoimmune Polyendocrinopathy Syndrome and its Unusual Presentation with Seizures

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Background: Fahr's syndrome is a rare neurological disorder, characterized by symmetric calcifications in basal ganglia, cerebellum, and cerebral cortex, secondary to genetic, infectious, and autoimmune etiologies which can lead to movement and gait disorders, cerebellar and speech abnormalities, and cognitive impairment. Fahr's disease is defined as primary familial idiopathic calcification, while Fahr's syndrome is caused by secondary factors that cause hypoparathyroidism. In this case report, an adolescent with a unique sequence of different manifestations of APS-1 is described who eventually developed Fahr's syndrome.

Case Description: Patient presented to us with abnormal body movements and painful spasms of limbs. Movements were in the form of twisting of arms and legs, associated with stiffness of hands and feet. He was diagnosed with IDDM at 10 months of age, had been on insulin with poor compliance. The child had intermittent twitching of his arms. He had diabetic arthropathy, mal-aligned teeth and oral thrush. Neurological examination was unremarkable. Labs showed Total calcium 5.9 mg/dl (8.6-10.2 mg/dl), phosphate 8.3 mg/dl (4.0-7.0 mg/dl), vitamin D3 7.0 ng/mL (30–100ng/dL), iPTH level 11.50 pg/ml (16–87 pg/ml), HbA1c 10.4%. Addison, thyroid and celiac screens were negative. EEG showed a single episode of generalized spike and slow waves with a slow posterior dominant rhythm. MRI brain with contrast showed bilateral symmetrical abnormal T1 hyperintense signals in basal ganglia, thalami, red nuclei, dentate nuclei and bilateral cerebellar folia. CT brain revealed symmetrical diffuse extensive calcification. Treatment focused on correcting hypocalcemia, managing seizures, controlling diabetes, and treating candidiasis. He showed significant improvement in symptoms with normalization of calcium and phosphorus. His genetic testing was planned for APS-1.

Conclusion: APS-1 manifests in early life and affects the endocrine and nonendocrine systems. This case highlights the importance of considering Fahr's Syndrome as a possible cause in patients with polyendocrinopathies having neurological symptoms.

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PITUITARY NEUROENDOCRINOLOGY, AND PUBERTY

A Case of Pediatric Female Central Diabetes Insipidus Presenting Severe Malnutrition and Stunting

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Background: Central diabetes insipidus is a syndrome characterized by polyuria and polydipsia, resulting from a deficiency of the arginine vasopressin hormone. This condition may arise due to defects in the synthesis or secretion processes within the neurons of the posterior pituitary gland. Chronic malnutrition is a significant complication resulting from persistent extreme thirst. While individuals with central diabetes insipidus can maintain a good quality of life, they also at risk of potentially life-threatening conditions due to imbalances in fluid and electrolytes.

Aim: To observe the long-term clinical outcomes of pediatric female patient with central diabetes insipidus who present with severe malnutrition and stunting that receives desmopressin therapy (a vasopressin analogue).

Case: A 2-year-and-11-month-old girl with central diabetes insipidus, diagnosed at 2 years and 6 months of age, received desmopressin (a vasopressin analogue) therapy. At the initial diagnosis, her diuresis ranged from 8.5 to 21.5 ml/kg/hour. Treatment with titrated doses of desmopressin showed gradual improvement in diuresis with controlled electrolytes profile and stable renal function over a 12-month period of observation. The patient's nutritional status improved slowly but was not yet optimal, progressing from severe malnutrition to mild-tomoderate malnutrition. Her height-for-age status improved from stunting to normal height-forage within 12 months of observation. Thirst remained extreme with her tendency to seek cold water, proving that there was no adipsia. There were no episodes of electrolytes imbalance, dehydration, and desmopressin-related side effects. A contrast-enhanced brain magnetic resonance imaging (MRI) revealed a normal condition of the pituitary gland and its surroundings structures.

Conclusion: The outcomes of pediatric female patient with central diabetes insipidus receiving long-term desmopressin therapy showed improvements in polyuria, polydipsia, nutritional status, and controlled electrolytes profile. Keywords: central diabetes insipidus, arginine vasopressin deficiency, malnutrition, desmopressin, vasopressin analogue

FSH and Sertoli cell biomarkers: The long-awaited potential answer to distinguishing hypogonadotropic hypogonadism from self-limited delayed puberty

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Background: Delayed puberty is a common concern in males and distinguishing between self-limited delayed puberty (SLDP) and congenital hypogonadotropic hypogonadism (CHH) is particularly challenging. Traditional endocrine tests, which typically measure stimulated luteinizing hormone (LH) or testosterone levels, often fail to provide a clear diagnosis. Since follicle-stimulating hormone (FSH) stimulates Sertoli cells, leading to testis enlargement and increased secretion of anti-Müllerian hormone (AMH) and inhibin B, and given that the FSH-Sertoli cell axis remains functional during normal childhood and early puberty, we aimed to explore whether measuring serum FSH, AMH, and inhibin B could help differentiate between SLDP and CHH.

Objective: To assess whether serum levels of FSH, AMH, and inhibin B can effectively distinguish between SLDP and CHH.

Study Design: We conducted a prospective, nested case-control study involving a cohort of male adolescents with delayed puberty. Baseline serum reproductive hormone levels were compared to identify potential biomarkers for CHH, with participants followed until a definitive diagnosis was confirmed using gold standard criteria (age 18 or ≥4 years after testis volume reached 4 mL).

Results: Among the 65 participants who completed the follow-up, 33 were diagnosed with SLDP and 32 with CHH. Serum FSH, AMH, and inhibin B demonstrated superior diagnostic accuracy compared to LH and testosterone in distinguishing between SLDP and CHH. A combination of FSH (IU/L) × inhibin B (ng/mL) <92 and FSH (IU/L) × AMH (pmol/L) <537 exhibited high sensitivity (>93%), specificity (≥92%), predictive values (>92%), and a positive likelihood ratio (>12) for diagnosing CHH.

Conclusions: Serum FSH combined with inhibin B or AMH is highly predictive to accurately distinguish between SLDP and CHH in adolescent males.

POSTER ABSTRACTS

SYNDROMES

Genetic variants in a group of patients with Noonan Syndrome in a pediatric hospital

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Introduction: Noonan Syndrome is caused by mutations in PTPN11 (12q24.13) in 50% of cases, SOS1 (2p22.1) in 15%, RAF1 (3p25.2), RIT1 (1q22) and LZTR1 (22q11.21), and less frequently in other genes associated with the RAS/MAPK signaling pathway. The clinical spectrum of SN may differ slightly depending on the causative genes, and “Noonan-like” forms (SN-like disorder with juvenile myelomonocytic leukemia and SN-like disorder with loose anagen hair) have also been described.Clinical features are short stature, dysmorphic facial features, congenital heart defects, most commonly pulmonary valve stenosis, typical chest, cryptorchidism.

Results: We describe 6 patients with RASopathy Noonan syndrome : 5 males, 1 female, with an average gestational age of 37 weeks, 2.9 kg of weight and 45.5 cm of height at birth. 100% had: palpebral ptosis, winged neck, pectum carinatum and short stature, 83% had heart disease such as subaortic stenosis and ventricular septal defect, and 16% had hypoacusis and altered genitalia. The genetic variants found in the PTPN 11 gene,(3male 1 female) all heterozygous were: in the sporadic males: Exon 7 c.836 A>G. pTYR:279cys, and c.417G>C (p.Glu139Asp) and p.Asn 308 Ser , c..923A>G heterozygosis . the female with variant in c.417G>C (p.Glu139Asp) whose mother has SN. One male with variant in KRAS c.64_65insCCT p.Gln22delInsProTer Het, variant showing AGG insertion at the 25245321 position on chromosome 12 , another male with a variant in RAF1p.ser257Pro otra c.769 T>C YOTRA G.12645700A>G en heterocigosis patogenic RAF protooncogenserine/threonineproteinkinasa NP-0002871.1:p.Ser257Pro/NC000003.11.:g.12645700A>G.table 1.

Analysis and conclusions: We found genetic alterations in PTPN11 and less frequently in other genes associated with the RAS/MAPK signaling pathway.It is essential to typify the clinical and genetic alteration in patients with RASopathies so that physicians involved in the care of these patients are familiar with the diagnosis, genetic variant, manifestations and clinical follow-up, especially because of their predisposition to malignancy.

Multinodular Goiter as a Presentation of DICER1 Syndrome

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DICER1 syndrome (D1S) is an autosomal dominant hereditary tumor predisposition syndrome that leads to the development of a variety of benign and malignant tumors during childhood and adolescence. The earliest reported tumor associated with this syndrome is pleuropulmonary blastoma. Early-onset multinodular goiter (MNG) and thyroid carcinomas have also been described.

Case reports: The clinical charts, laboratory tests, treatments, and outcomes of six girls (age range: 10–14 years) with MNG were retrospectively reviewed. Five out of six patients had first-degree relatives with MNG or papillary thyroid cancer, and two of these five had mothers and aunts with benign ovarian cysts. The patient without a family history of MNG underwent unilateral oophorectomy at the age of 12 due to an ovarian tumor (adult-type granulosa cell tumor), while another patient had a history of Wilms tumor. All patients had normal thyroid function and negative thyroid antibodies. Thyroid ultrasound (US) revealed multiple thyroid nodules in all patients at the time of diagnosis. Fine-needle aspiration biopsy (FNAB) of predominant and/or suspicious thyroid nodules (TN) was performed on all patients, revealing benign cytology (Bethesda II) in five out of six and non-diagnostic results (Bethesda I) in one case. Following a close follow-up period (8–24 months), total thyroidectomy was indicated in four out of six cases: three of the four due to suspicious US findings (three out of five with growing TN, one out of five with persistent BMN) and two cases due to a mutual decision between medical professionals, patients, and their parents. Histology revealed diffuse nodular hyperplasia in three out of six cases (one of which had atypical follicular adenomas) and papillary thyroid carcinoma in two out of five (both ATA low risk). Given the family or personal history of thyroid and/or gynecological tumors, genetic testing for the DICER1 gene was conducted, and pathogenic variants were identified in all patients. Following the diagnosis, gynecological ultrasounds were requested in five out of six patients (one was already under gynecological follow-up).

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Two of these patients were found to have benign ovarian tumors and underwent tumor excision: one with a moderately differentiated Sertoli-Leydig cell tumor and the other with a well-differentiated mucinous cystadenoma.

Conclusion: A positive personal or family history of MNG or gynecological tumors in a patient presenting with MNG should raise suspicion for a DICER1 gene mutation. As this syndrome can be associated with either benign or malignant thyroid tumors during childhood or adolescence, its diagnosis allows for early detection of associated pathologies. Given the current understanding of the generally benign prognosis of this condition, the risks versus benefits of lifelong screening must be carefully considered. Nevertheless, reporting such cases will contribute valuable information toward the ongoing refinement of care guidelines.

Turner Syndrome: When Should We Suspect it?

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Introduction: Turner syndrome (TS) results from a partial or complete monosomy of the X chromosome, with an incidence of approximately 1 in 2500 live female births. Clinically, TS is characterized by short stature, gonadal dysgenesis, and a distinct phenotype. It is often associated with various comorbidities, including cardiovascular, renal, hepatic, autoimmune disorders, hearing impairment, and neurocognitive anomalies.

Objectives: This study aims to present a case of a patient with TS whose clinical presentation was atypical.

Case: A 15-year-old female patient was referred to our service for evaluation of short stature. She exhibited a height of 122 cm (-6.06 SD), a weight within the 25th to 50th percentiles, and significant body disproportion (percentile >97). Physical examination revealed facial puffiness, cold and dry skin, vulvar vitiligo, and a pilomatrixoma on her back. The neck appeared thickened, the thyroid gland did not show signs of enlargement, and edema was noted in both upper and lower extremities. The patient was assessed to be prepubertal. Laboratory investigations demonstrated severe hypothyroidism, elevated liver transaminases, dyslipidemia, and hypergonadotropic hypogonadism (Table 1).

	Resultado	Valor de referencia
GOT/GPT (UI/L)	183/309	<33
Colesterol (mg/dl)	225	<170
HDL/LDL (mg/dl)	40/157	>45/<110
Triglicéridos (mg/dl)	139	40-130
Ac Anti transglutaminasa (U)	2,9 (neg.)	<20
PRL (ng/ml)	669	3-25
TSH (uU/ml)	>100	0,5-6,5
T4L (ng/dl)	0,64	0,8-2,2
ATPO (UI/ml)	>600	<20
ATG (UI/ml)	181	<20
LH (mU/ml)	19,53	0,10-1,60
FSH (mU/ml)	76,91	0,60-4,60
E2 (pg/ml)	<10	<10-20
AMH (pmol/l)	<1,2	5-55

Thyroid ultrasound indicated a heterogeneous, diffusely micronodular gland with a solid nodule measuring 8x8 mm and exhibiting central vascularization in the left lobe. An echocardiogram revealed mild tricuspid regurgitation and an anomalous tendon in the left ventricle. Gynecological ultrasound showed a prepubertal uterus with absent ovaries. Bone age assessment indicated a delay corresponding to 10 years, with findings of triangularization of the distal radial epiphysis and pyramidalization of the carpal bones, suggestive of SHOX gene alteration. Treatment with levothyroxine was initiated. Given the severe short stature and the radiological findings, TS was strongly suspected, and this diagnosis was subsequently confirmed through karyotyping, which revealed a 45,X chromosomal pattern.

Conclusions: The patient presented with a clinical picture consistent with severe hypothyroidism, and the combination of clinical and radiological findings led to a suspicion of TS. Recognizing the distinctive features of this syndrome is crucial for timely diagnosis and intervention.

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Prevalence of Cardiovascular Abnormalities in Children and Adolescents with Hyperthyroidism at the Lagos University Teaching Hospital-A Cross-Sectional Study

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Background: Hyperthyroidism is a rare but serious disorder in childhood occurring most frequently as a consequence of Graves' disease (GD), an autoimmune disorder resulting from thyrotropin (TSH) receptor stimulation by autoantibodies. Graves' disease is the most common cause of hyperthyroidism in pediatric population accounting for 10–15% of thyroid diseases in children younger than 18 years. In our previous study (8years ago) of pattern of thyroid disorders seen in children in the Lagos University Teaching Hospital (LUTH), Nigeria over a 10-year-period, GD constituted 12.7% of the total children with thyroid disorders. Previous studies involving largely adult patients with GD indicated that incidence of thyrotoxic heart disease in sub-Saharan Africa range from 6.2-27% and that thyrotoxicosis is a notable cause of cardiac morbidity. Studies from North Africa and Asia have reported cardiovascular dysfunction in children with GD. There is paucity of studies on cardiovascular dysfunction in children with hyperthyroidism in Sub-Saharan Africa with the only study found being a recent conference poster presentation on a case series of 4 adolescents from South-South Nigeria.

Objective: To evaluate cardiovascular functions in children and adolescents with hyperthyroidism attending the paediatric outpatient clinic of LUTH.

Methods: A cross-sectional study involving patients aged from 0 to 18 years diagnosed with previous or new-onset hyperthyroidism was carried out after obtaining ethical approval from the LUTH health research ethics committee. Exclusion criteria included known prior cardiovascular disease before onset of hyperthyroidism or congenital heart disease. Written informed consent and assent were obtained from parents and patients respectively before recruitment. Available clinical notes and investigations were retrieved from participants' files. Cardiovascular functions were assessed by electrocardiography and echocardiography. Data analysis was done with Statistical Package for Social Scientists (SSPS) version 25.

Results: Nineteen children and adolescents were recruited. There were 3 males and 16 females giving a male: female ratio of 1:5. The mean (SD) age at presentation was 10.8±4.3 years while the mean (SD) present age of participants was 14.2±4.3years. The median (range) duration of disease was 3 (1-9) years. Eleven participants (57.9%) could afford to do TSH receptors antibodies at diagnosis and the titres were elevated except in one patient. None of the patients had achieved remission. Two participants had ECG abnormalities of borderline first degree heart block and left axis deviation respectively. Fourteen (73.7%) participants had mild to moderate regurgitation of the tricuspid, pulmonary, mitral and aortic valves while four (21%) participants had significant dilation of the cardiac chambers. Three (15.8%) participants had combined chamber dilatation and valvular lesions and the duration of disease in these was two, three and nine years respectively.

Conclusion: A high prevalence of valvular regurgitation and cardiac chamber dilation was noted among study participants. High rate of non-remission of GD was seen. Regular monitoring of cardiovascular functions in pediatric populations with GD is advocated. More definitive treatment of GD in patients not achieving remission by surgery or radioactive Iodine treatment (in appropriate age group) may help to prevent further cardiac decompensation and increased morbidity and mortality.

Growth and Neurodevelopmental Outcomes of Children with Congenital Hypothyroidism Identified Through Newborn Screening at a Tertiary Care Hospital in Pakistan

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Background: Congenital Hypothyroidism stands as the most common congenital endocrine disorder in childhood and a leading preventable cause of mental retardation. Despite its prevalence, global newborn detection rates remain suboptimal. The main hurdle in implementing newborn screening in Pakistan is lack of healthcare infrastructure coupled with lack of prevalence data and economic crises. The Aga Khan University Hospital spearheaded congenital hypothyroidism screening in Pakistan in March 1987, followed by dried blood spot newborn screening in April 2019. The estimated CH incidence in Pakistan ranges from 1:1000 to 1:1600, which is much higher than the worldwide incidence of 1:2000-3000.

Objective: To determine the growth and neurodevelopmental outcomes of children with primary congenital hypothyroidism identified through DBS newborn screening program in Aga Khan University Hospital over five years from April 2019 to April 2024.

Methods: Retrospective review of data from medical records of all confirmed congenital hypothyroidism cases was done. Data including the demographics, antenatal and family history, clinical features, age at diagnosis and start of treatment, initial TSH, FT4 levels, thyroxine dose increment at one year, follow-ups in the clinic, growth and neurodevelopmental outcomes were studied. Data was recorded on a structured proforma and identity was kept anonymous.

Results: 41 babies were diagnosed with congenital hypothyroidism. Incidence rate was 1 in 1000 live births. Out of all, 58% were females, 7 babies had a birth weight 33 weeks. 8 mothers had hypothyroidism. DBS TSH was >20 in 27 babies (65.8%), serum TSH was >100 in 16 babies (39%) with depressed FT4 in 36.5%. Mean age at diagnosis was 8.4 days while at treatment initiation was 10.8 days. 8 babies were symptomatic at the time of diagnosis with prolonged jaundice being the most common symptom. 2 babies had associated congenital nephrotic syndrome, out of which one baby expired due to complications of nephrotic syndrome, one had trisomy 21, and one had imperforate anus. Follow-up showed increments in doses at 1 year in majority (90%), 1 child had isolated speech delay, 1 had developmental delay requiring neurodevelopmental evaluation while the growth of the majority (95%) of babies was normal.

Conclusion: Incidence of CH was reported 1:1000 surpassing the global incidence, highlighting the urgency for early detection and treatment strategies at the government level. Most babies were asymptomatic at the time of diagnosis, making screening a mandatory tool to prevent adverse long-term outcomes.

Congenital Hypothyroidy in an Adolescent Diabetic in Kinshasa. Diagnosis Difficulty in an Under-equipped Environment

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Introductory: Congenital hypothyroidism is a pathology for which screening is systematically recommended in the neonatal period. Among the causes is congenital thyroid dysgenesis including athyreosis which is one of the most severe forms. The evolution depends on the clinical form, the intensity of the hormonal deficiency and the duration of treatment. In developed countries, the prognosis of children has been significantly improved thanks to access to diagnostic means and early treatment. In developing countries, particularly in the Democratic Republic of Congo, publications on this subject are non-existent.

We present a case of congenital hypothyroidism of the athyreosis type in a diabetic adolescent in Kinshasa, a form rarely described in sub-Saharan Africa, emphasizing the diagnostic difficulties in an under-equipped environment.

Patient and Observation: This is a 14-year-old girl, known to be diabetic and followed since the age of 7, under insulin therapy. The medical history indicates a low birth weight of 2100 grams (below percentile 10), hospitalization in a context of jaundice, constipation and fatigue. The current physical examination shows macroglossia, failure to thrive (height 151 cm, weight 30 kg), BMI of 13.6 kg/m2, delayed puberty, significant intellectual deficit and hypoacusis. Cervical ultrasound showed empty hyperechogenic structures in the thyroid compartments. The average level of thyroid-stimulating hormone “TSH” was 38 mIU/L (VN 0.5 – 5.5 mIU/L) and the level of free thyroxine was 6 pmol/m (VN 12-18 pmol/l) , the transcription factors FOXE1, NKX2-1 NKX-5 or PAX8 (9q22, 14q13, 5q34 and 2q12-q14) were not analyzed. We made the diagnosis of congenital hypothyroidism of the athyreosis type. Therefore, substitution treatment with Levothyroxine was started at a dose of 400 µg or 13.3 µg/Kg with quarterly monitoring.

Conclusion: We presented the first case of congenital hypothyroidism of the athyreosis type in the Democratic Republic of Congo with a poor clinical course, due to lack of diagnosis and early treatment. The diagnosis was incomplete, genetic analyzes inaccessible. This case illustrates the diagnostic and therapeutic difficulties as well as the need for systematic screening of congenital hypothyroidism in our environment

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Acute Suppurative Thyroiditis

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Lumps around the thyroid gland in children are often associated with changes in thyroid function, as seen in cases of hyperthyroidism and hypothyroidism. Infection is one of the frequently overlooked causes of lumps in the front neck area. Acute suppurative thyroiditis is a rare finding in pediatric infections that can develop into a thyroid abscess. This condition is rare, accounting for 0.1-0.7% of thyroid cases. This disease is associated with local infections such as dental caries. A study by She in 2022 showed that 2 out of 18 study samples also had dental caries in addition to acute suppurative thyroiditis but further research is needed to confirm the connection between these conditions.

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Case Report: This is a case report of a 7-year-old girl who presented to the pediatric endocrinology clinic with complaints of a lump approximately 4-5 cm in diameter on the left side of the front of her neck, which appeared about a month prior to her visit, accompanied by fever and pain. The patient also complained of toothache and swollen gums before the lump appeared. Ultrasound (USG), laboratory tests, MRI, and dental examinations were performed to establish a diagnosis. USG findings suggested suspected malignancy with a differential diagnosis of abscess and lymphadenitis. Laboratory tests showed elevated leukocytes, ESR, banded and segmented neutrophils, and lymphocytes, which were consistent with an infection. The patient's TSH and FT4 levels were normal. The dental examination revealed multiple tooth infections, requiring extractions. An ultrasound-guided FNAB was performed for further investigation, revealing no signs of malignancy. MRI of the neck and thyroid with contrast showed no enlargement of the left or right thyroid and identified a diffuse inhomogeneous lesion obliterating the left thyroid parenchyma, leading to a diagnosis of acute suppurative thyroiditis on the left side, without lymphadenopathy. The patient received antibiotic therapy and treatment for the dental infection. After antibiotic treatment, the neck lump reduced to a diameter of 1-2 cm, and the pain subsided, though the lump had not completely disappeared. A biopsy was planned if antibiotic therapy failed, but the patient's family refused the procedure.

Conclusion: Acute suppurative thyroiditis is an entity which is not seen much in routine but can cause significant morbidity. Similar symptoms can pose a diagnostic challenge, as they overlap with other thyroid diseases such as hyperthyroidism, hypothyroidism, or thyroid cancer. Early diagnosis is possible with clinical suspicion, pathological and imaging investigations (USG and MRI with contrast) can help make early and proper diagnosis, thus helping clinicians to start timely treatment.

Case Report and Insight into the Congenital Hypothyroidism in Myanmar

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Congenital Hypothyroidism (CH)is the critical Paediatric Endocrine Disorder that can lead to permanent developmental delayed if it's not timely treated. In Myanmar, lack of neonatal screening, the challenges of health care access contributes to delayed recognition , diagnosis , and management of CH. , The case reports explores the situation of congenital hypothyroidism in Myanmar.

Case 1 -- 14 months old baby girl came to our Pediatric and Endocrine clinic for delayed developmental milestone The girl was born normal vaginal delivery, birth weight was 3kg and had history of prolonged neonatal jaundice , constipated. The baby exhibited signs consistent with congenital hypothyroidism like coarse facial appearance, lethargic , distended abdomen , hypotonia. The TSH level was >100uIU/ml, Free T4 1.07 pmol/L , free T3 100uIU/ml , free T4 9.17 pmol/L , free T3 5.19 pmol/L .

Case 2 -- 5months old baby girl was referred to our Pediatric Endocrine Clinic. She was born normal delivery at home in rural area, birth weight was 3.5 kg The parents noticed prolonged neonatal jaundice . But they thought it was normal for the newborn. The local pediatrician referred her to our clinic. The girl couldn't control head, and had myxedema face , pallor , dry skin , constipation were noticed . TSH was >100uIU/ml , free T4 9.17 pmol/L , free T3 5.19 pmol/L .

We have many infants who got early diagnosis and proper treatment of CH and have normal growth and development. But the above cases highlighted critical challenges of the diagnosis and management of CH in Myanmar. The Pediatric Endocrine and Diabetes clinic was started at Yangon Children Hospital in 2014. It's first Pediatric and Endocrine clinic in Myanmar. We increased awareness of the CH among health care workers to have early diagnosis of the CH. At our clinic we have more than 150 cases of CH , less than quarter of the cases were diagnosed after neonate and some few cases diagnosed after infancy. Most of the cases came to our clinic due to referral from the local doctors or pediatricians. The management of the CH is still challenging since Covid Pandemic and crisis in Myanmar. The cost , availability and reliability of thyroid function tests varies significantly depending on the location and infrastructure .The high cost deter early screening and to repeat the tests to confirm or monitor. The crisis has exacerbated the situation.

Globally, newborn screening for CH is a standard practice. In contract, Myanmar faces challenges due to the lack of recourses to implement comprehensive newborn screening , limited access to health care facilities , insufficiency public awareness , socioeconomic barrier and unstable situation. By overcoming these challenges, Myanmar can improve the early detection and management of CH . There by preventing the long-term cognitive and developmental improvement and public health improvement. We continue our efforts to improve CH care in Myanmar.