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Poster Presentations

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Circulating maternal microRNAs and preterm birth in South African women: impact of human immunodeficiency virus and antiretroviral therapy

Category: Basic Science

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Background: Preterm birth (PTB) is the leading cause of neonatal mortality and increases the risk of cardiometabolic disease later in life. PTB is also more common in women living with human immunodeficiency virus (HIV). Circulating maternal microRNAs (miRNAs) during pregnancy hold promise as biomarkers for PTB, but there is limited data on the impact of HIV infection and antiretroviral therapy (ART) in South Africa where HIV infection is common. This study aimed to identify miRNAs associated with PTB and assess the impact of HIV status and ART regimen in South African pregnant women.

Methods: This nested case-control study included 76 pregnant women, matched by age and body mass index (BMI), comprising 38 with PTB and 38 with term births. Participants included women living without (n = 28) and women living with HIV receiving either dolutegravir (n = 24) or efavirenz combination therapies (n = 24). MiRNAs (n = 179) were profiled in serum collected between 24 and 28 weeks of gestation using miRNA PCR array panels.

Results: MiR-320d, miR-29b-3p, miR-22-5p and miR-30e-5p were associated with PTB in women without HIV and those receiving efavirenz ART with receiver operating characteristic area under the curve (AUC) values ranging from 0.85 to 0.92. These miRNA were not associated with PTB in women receiving dolutegravir ART however, where only miR-154-5p was associated with PTB (AUC 0.86).

Conclusion: In conclusion, our findings highlight that HIV/ART may impact the use of miRNAs as biomarkers for PTB, offering valuable insights for biomarker discovery in populations with high HIV prevalence.

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Comparing the influence of regularly prescribed drugs on the chemotactic effects of autologous blood clots on PBMC migration in type 2 diabetic patients

Category: Basic Science

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Background: Impaired wound healing is a major complication of diabetes mellitus. Topical autologous blood clot therapy (TABCT) is an autologous scaffold used to mimic the initial haemostasis response and stimulate healing. It is, however, unclear if this therapeutic approach would be effective in diabetic wounds given the multifactorial immune dysfunction evident in these patients. This study compared the effect of regularly prescribed drugs (prednisone, amoxicillin, rifampicin and metformin) on chemokine release and subsequent immune cell chemotaxis in the context of TABCT.

Methods: Twelve participants were recruited and subdivided into healthy controls (n = 6) and T2DM (n = 6) patients. Standard anthropometric measures and blood glucose were assessed. Whole blood was "spiked-in" with pharmaceutical drugs of interest before inducing clot formation ex vivo. The release of CCL5 from the blood clots was determined using an ELISA assay. Subsequently, the chemotactic effect of the clot-secretome on peripheral blood mononuclear cell (PBMC) migration was assessed using live cell imaging.

Results: Prednisone increased the size of blood clots as well as CCL5 release in both healthy controls and T2DM patients but did not affect PBMC migration. Compared to standard culture conditions, the clot secretome decreased the velocity and distance of PBMCs during migration but increased their directionality, confirming its chemotactic effect. Rifampicin diversely affected the clot-induced PBMC migration (velocity, distance) in control compared to T2DM patients.

Conclusion: This study expanded on the effect of pharmaceuticals on TABCT.

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Glycaemic control mechanisms of Litchi chinensis Sonn. (Litchi) peel ethyl acetate extract in a fructose/streptozotocin diabetic model of rats

Category: Basic Science

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Background: The glycaemic control potential and flavonoid profile of litchi have been documented for its hydroalcoholic extracts, while there is scarce information regarding its ethyl acetate extract. This study investigated the flavonoid profile, as well as the ameliorative potential and possible underlying mechanisms of litchi peel ethyl acetate extract on type 2 diabetes-related pathologies in a fructose/streptozotocin (STZ) model of diabetic rats.

Methods: Sprague Dawley rats were induced with diabetes by administering 10% fructose for 2 weeks and a single i.p. injection of low-dose (40 mg/kg bw) STZ. Thereafter, the animals were orally treated with a low-dose (150 mg/kg bw) and high-dose (300 mg/kg bw) of the extract (LDPE and HDPE, respectively) and metformin (200 mg/kg bw). Various diabetes-related indices were assessed in the animals and their biological samples. The notable flavonoids in the extract determined using LC-MS.

Results: Compared to untreated diabetic rats (AUC = 1004 mg.h/dL), the HDPE significantly ($p < 0.05$) improved glucose tolerance (AUC = 847 mg.h/dL), which was statistically comparable ($p > 0.05$) to the effect of metformin (AUC = 903 mg.h/dL). Serum insulin and pancreatic histology data showed that the STZ-induced pancreatic damage and insulin depletion was improved by the HDPE, which could be linked to the observed ameliorative effect of the extract on pancreatic lipid peroxidation and SOD and catalase activity. The extract further improved liver and muscle glycogen storage, as well as muscle hexokinase activity and Akt phosphorylation, suggesting that the extract exerts glycaemic control by enhancing glycogen storage and modulating insulin-mediated signalling of glucose uptake and utilisation. LC-MS data and documented reports suggest that flavonoids, such as epicatechin, cinnamtannin B2, procyanidin B5, and proanthocyanidin A2, are the possible influencing compounds.

Conclusion: The ethyl acetate extract of litchi peel could be a source of bioactive flavonoids that can potentiate glycaemic control in diabetes and mitigate oxidative stress-related pathologies.

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Evaluation of a point-of-care testing platform against a HPLC HbA1c measurement method in a South African tertiary-level diabetes clinic

Category: Basic Science

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Background: Diabetes mellitus (DM) is a global issue, with South Africa having the highest sub-Saharan African prevalence (11.3%). Limited access to glycated haemoglobin(HbA1c) testing delays treatment adjustments for diabetic patients. Point-of-care (POC) analysers provide faster results, potentially enhancing diabetes management. This study aimed to evaluate the accuracy of a POC device compared to a laboratory-based method.

Methods: A prospective cross-sectional study conducted at Charlotte Maxeke Johannesburg Academic Hospital over 4 months, analysed whole blood and capillary samples from diabetic patients using the Finecare™ HbA1c Rapid Quantitative POC device. These results were compared to the Bio-Rad Variant II, a laboratory method utilising ion-exchange high-performance liquid chromatography (HPLC). Haemoglobin (Hb) variants identified from routine laboratory HPLC analysis were also assessed on the POC device. Accuracy was evaluated against IFCC standards (Total Allowable Error = 6.9%).

Results: The POC device demonstrated a strong correlation with HPLC for both capillary (r = 0.94, 95% CI 0.90–0.96) and whole blood remnant samples (r = 0.94, 95% CI 0.91–0.97). Capillary and whole blood remnant samples also correlated well (r = 0.95, 95% CI 0.93–0.97). Mean bias was 1.3% (95% CI -1.2–3.8, p = 0.17) for capillary vs. HPLC, 4.5% (95% CI 2.1–6.9, p = 0.0003) for whole blood remnant vs. HPLC, and 3.3% (95% CI 0.9–5.6, p = 0.01) between capillary and whole blood remnant samples. However, POC whole blood remnant samples with Hb variants showed poor correlation with HPLC (r = 0.60, 95% CI 0.37–0.77) and a mean bias of -2.8% (95% CI -7.2–1.5, p=0.2).

Conclusion: The Finecare™ HbA1c POC device performed acceptably with both sample types, comparable to the HPLC method. We recommend capillary samples for routine monitoring but advise caution in populations with high Hb variants.

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The novel role of long non-coding ribonucleic acids at the pre-diabetes stage in multi-ethnic patients aged from 25–45 years from Durban, South Africa

Category: Basic Science

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Background: Pre-diabetes is a condition that precedes the onset of type 2 diabetes (T2D) where the body has high blood glucose levels but not high enough for T2D diagnosis. Long non-coding ribonucleic acids (LncRNAs) have been reported to be expressed in immune cells such as non-coding transcript in T- cells (LncRNA-NTT), non-coding Repressor of nuclear factor activated T-cells (LncRNA-NRON), and non-coding RNA expressed in dendric cells (LncRNA-DC). In this study, we sought to investigate the expression of selected LncRNAs at the pre-diabetes stage in multi-ethnic population aged from 25–45 years from Durban (South Africa).

Methods: Upon ethical approval, samples ($n = 46$) were collected from King Edward Hospital. They were categorised into 3 groups which are, non-diabetic (ND) ($n = 9$), pre-diabetic (PD) ($n = 15$) and type 2 diabetic ($n = 22$). LncRNAs relative expression were measured using qPCR and data analysed using SPSS statistics v28.

Results: Results showed a statistically significant ($p < 0.05$) increase in LncRNA-NRON, an increase in LncRNA-DC expression and a decrease in expression of LncRNA-NTT expression in PD group by comparison with ND group.

Conclusion: LncRNAs expression reported during immune response paves the way for exploration of immunity at genetic level using immune cells. The results obtained in this study gave pivotal insights into the expression of these LncRNAs during pre-diabetes state. Further investigation would be very useful as this activation involve certain cells such as macrophages and dendric cells which have not yet been measured at PD stage and also the measurement of other inflammatory cytokines such as IL-2 and type I interferons.

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The prevalence, aetiology and healing trajectories of hard-to-heal wounds in South Africa

Category: Basic Science

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Background: The incidence of hard-to-heal wounds is rising globally with adverse effects on quality of life. Yet, there is no reliable data available on hard-to-heal wound prevalence, aetiology, and outcomes in a low-to-middle income country without improper care being a confounding factor.

Methods: A retrospective study of 460 individuals (876 wounds) that received appropriate standard of care at a specialised wound care clinic in the KwaZulu-Natal province of South Africa.

Results: Acute/traumatic wounds were most prevalent (230/460, 50%) followed by ulcers (173/460, 38%) (DFUs 13%, VLUs 12%, PIs 11%, MLU < 1%, ALUs < 1%) and atypical wounds (55/460, 12%) (atypical wounds 8%, vectors 4%). Delayed referral for specialised wound care was evident for individuals with ulcers. One hundred and three out of 460 (22%) individuals did not respond to standard of care and were classified as hard-to-heal (12 weeks of standard of care). Diabetes mellitus (45/103, 44%) and wound infection (44/103, 43%) accounted for poor healing trajectories in the hard-to-heal cohort, whereas 14/103 (13%) individuals had other comorbidities.

Conclusion: High prevalence rates of hard-to-heal wounds in the heterogenous South African population necessitate recognition of wound management as a specialty in South Africa.

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A case of acromegaly first diagnosed in pregnancy, at Chris Hani Baragwanath Academic Hospital

Category: Clinical

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Introduction: Acromegaly is a rare disorder caused by excessive growth hormone (GH) production, usually due to a pituitary adenoma. As a result of the associated hypogonadotropic hypogonadism, spontaneous conception is uncommon, and pregnancy is rare. Diagnosis during pregnancy is challenging, as physiological changes can mask symptoms, and management poses unique therapeutic dilemmas.

History and examination findings: We present a case of a 31-year-old female referred to the Gestational-Endocrine Clinic at 34 weeks' gestation with gestational diabetes. Further evaluation revealed features suggestive of acromegaly, leading to biochemical and radiological confirmation.

Investigations: Despite the late diagnosis, the patient had a positive pregnancy outcome with no immediate obstetric complications. Postpartum, she underwent successful transsphenoidal surgery for pituitary adenoma resection, followed by medical therapy for residual disease. Long-term follow-up focused on endocrine control, metabolic complications, and tumour surveillance.

Conclusion: This case highlights the diagnostic complexities of acromegaly in pregnancy, the interplay between pregnancy and pituitary tumour behaviour, and the importance of a multidisciplinary approach for optimal maternal and foetal outcomes. It underscores the need for heightened clinical suspicion in atypical presentations of gestational diabetes and hormonal disorders during pregnancy.

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Rare case of levothyroxine overdose

Category: Clinical

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Introduction: Levothyroxine is crucial for treating hypothyroidism but can be life-threatening when overdosed. There are limited cases reported, most of which are in children. Overdose, though rare, may lead to seizures, arrhythmias and thyroid storm. Despite its potential for clinical catastrophe, it is underreported, and management guidelines are scarce. Herein we report a case of levothyroxine overdose.

History and examination findings: A 36-year-old African male with retroviral disease, mitral valve insufficiency and Graves disease post-radioactive iodine therapy, ingested 10,000 µg of levothyroxine (~100 x 100 µg tablets) and 500 mg of warfarin (~100 x 5 mg tablets) in a self-harm attempt. He was stable on arrival, showing no signs of thyrotoxicosis or bleeding. Investigations revealed a fT4 > 100 pmol/L (reference range: 11.9–21.6 pmol/L) and an INR of 10.61. Vitamin K was administered for warfarin toxicity and levothyroxine was withheld. He was monitored in high care and received psychological therapy.

Investigations: Levothyroxine overdose in adults can be potentially fatal. While doses exceeding 5 000 µg can trigger thyrotoxicosis, endogenous compensatory mechanisms, such as increased reverse T3 production, can delay toxicity. Overdose should be suspected when T4 is markedly elevated with a normal TSH. Management includes β-blockers and glucocorticoids to inhibit peripheral T4–T3 conversion, with cholestyramine administered for severe cases. Activated charcoal and anti-thyroid drugs are ineffective, while extra-corporeal methods remain as salvage options.

Conclusion: This case highlights the critical need for close monitoring, even in the absence of acute symptoms, to prevent late-onset fatalities. A reminder that overdose can turn a hormone of healing into a catalyst of crisis.

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The prevalence of metabolic syndrome amongst staff members at Helen Joseph Tertiary Hospital, Johannesburg, South Africa

Category: Clinical

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Background: Metabolic Syndrome (MetS) is a cluster of conditions—including abdominal obesity, insulin resistance, hypertension, and dyslipidaemia—that significantly increase the risk of cardiovascular disease and type 2 diabetes mellitus (T2DM). The 2005 NCEP ATP III definition remains the most widely used classification for MetS. This study aimed to assess the prevalence of MetS among staff at Helen Joseph Tertiary Hospital (HJH).

Methods: A retrospective analysis was conducted on data from 220 HJH staff who voluntarily participated in screenings during World Diabetes Day 2024. Data collected included comorbidities, current medications, anthropometrics, HbA1c, and a lipogram. MetS was diagnosed in participants meeting at least three of the five NCEP criteria. A limitation of this study was the recording of random instead of fasting glucose due to the nature of the screenings. Therefore, the prevalence of MetS was calculated using known diabetics and those with an HbA1c ≥ 6.5%.

Results: The prevalence of MetS was 39.5%, with a female predominance of 88.9%. Increased waist circumference was observed in 50% of participants, while 62% had elevated blood pressure. Low high-density lipoprotein cholesterol was noted in 49% and 33% had high triglyceride levels. Additionally, 15.9% were either known diabetics (6.8%) or had an HbA1c ≥ 6.5% (9.1%) and were referred for further testing.

Conclusion: The elevated prevalence of MetS compared to other South African studies highlights the need for enhanced staff screening policies and health education initiatives alongside targeted interventions to prevent the progression to T2DM and cardiovascular disease.

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Advancing diabetic retinopathy detection: evaluating digital confocal photography at a tertiary diabetic clinic

Category: Clinical

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Background: Diabetes mellitus (DM) is highly prevalent in South Africa, and diabetic retinopathy (DR) is a leading cause of preventable blindness, highlighting the need for effective screening methods. This study aimed to assess the prevalence and severity of DR using advanced retinal imaging, while exploring its association with clinical and biochemical parameters.

Methods: A retrospective clinical audit was conducted on type 1 and type 2 diabetic patients at Charlotte Maxeke Johannesburg Academic Hospital, between April and May, 2023. Retinal photographs using the DRS Plus True Color Confocal Fundus

Scanner were captured and independently evaluated by two ophthalmologists.

Results: Of the 99 screened diabetic patients, the prevalence of DR was 33.7%. Patients with DR were significantly older than those without (54 vs. 42 years, $p = 0.005$), with the highest prevalence observed in those with T2DM (36.5%). A longer duration of T2DM (> 10 years) correlated with a higher DR rate. Coexistence of hypertension with DM increased the DR rate to 47%. Notably, 30% of individuals with DR reported no visual symptoms. Patients with DR had more microvascular complications compared to those without (53.6% vs. 46.4%, $p = 0.007$). While glycated haemoglobin (HbA1c) alone was ineffective for predicting DR (AUC 58.7), a model incorporating elevated systolic blood pressure, HbA1c, and dyslipidaemia showed 84% accuracy (AUC 0.88). Inter-observer reliability for DR reporting showed substantial agreement ($k = 0.61$, sensitivity 90%, specificity 76%, Accuracy 0.81).

Conclusion: Over one-third of diabetic patients had DR, with many presenting without visual symptoms, emphasising the need for comprehensive DR screening and management.

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Optimising CGM utilisation: collaborative in-person workshop for improving diabetes care

Category: Clinical

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Background: The effective use of continuous glucose monitoring (CGM) requires training in data interpretation, therapy adjustment and good engagement between healthcare professionals (HCPs) and people with diabetes (PWDs). This is especially important in resource-limited settings, where diabetes education is scarce and consultation time short. To meet these needs, we implemented two workshops for HCPs and PWDs in Cape Town and Johannesburg.

Methods: The 1-day workshops featured parallel and interactive sessions for PWDs and HCPs. HCP sessions covered best practices for insulin therapy, glucose profile analysis, and psychosocial aspects of care. The PWD sessions focused on optimising consultations with CGM data, managing stress and illness, and the impact of exercise and healthy living. Afternoon sessions included interactive panels and case studies. Feedback was collected to assess workshop effectiveness.

Results: A total of 103 participants (52 in Cape Town, 51 in Johannesburg) attended, equally representing PWDs and HCPs. Among all PWDs, 68% were female, 67% had type 1 diabetes, and 5% had type 2 diabetes. HCPs included 40% diabetes specialist nurses, 30% doctors, and 22% dieticians. Across all the participants, the Cape Town workshop increased CGM knowledge by 42% and confidence in using CGMs by 68%. In Johannesburg there was a 188% increase CGM understanding, with 52% gaining confidence in routine care.

Conclusion: The co-creation approach, combining diverse experiences with interactive sessions, effectively bridged knowledge gaps and boosted confidence in CGM use for diabetes care. This approach is replicable and can empower a community led development in CGM education and adaptation.

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Assessing the burden of cardiovascular risk at the University of the Witwatersrand: a hypertension focus

Category: Clinical

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Background: Non-communicable diseases are a leading cause of mortality in South Africa. The International Society of Hypertension initiated the May Measurement Month (MMM) in 2017 to screen and raise awareness regarding hypertension. This sub-study evaluates the incidence, prevalence, and risk-factor profile of newly diagnosed and uncontrolled hypertension, along with associated cardiometabolic risk factors among an educated cohort of students and staff at the University of the Witwatersrand.

Methods: A retrospective descriptive analysis was conducted using data from the MMM primary study, which included participants aged ≥ 18 years who were screened over four years (2019 and 2021–2023).

Results: Among the 1 749 individuals screened, 78% were aged 20–40 (mean age 30±13 years), and were predominantly female (63%) and black African. The overall prevalence of hypertension was 7.0% ($n = 122$) over the four years; 64.8% were newly diagnosed, and 35.2% were known hypertensives on pharmacotherapy (20.5% uncontrolled, 14.8% controlled). The prevalence of self-reported diabetes mellitus ranged from 1.5% to 5%. Nearly half of the participants were classified as overweight or obese in 2019; 36% were middle-aged (40–59 years), 29% were young adults (25–39 years), and 25% were youth (14–24 years). Sub-analysis by year, showed that being

overweight/obese was positively associated with hypertension but did not achieve statistical significance. Individuals in the middle-age group showed increased odds of hypertension (OR = 4.07, 95% CI: 1.69–9.81).

Conclusion: This study highlights hypertension›s heterogeneity, with higher cardiovascular risk factors in middle-aged individuals and greater alcohol and smoking prevalence among younger individuals, emphasising the need for targeted public health interventions.

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Prevalence of diabetic kidney disease in people living with type 2 diabetes mellitus at a tertiary level hospital in Johannesburg

Category: Clinical

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Background: Diabetes mellitus (DM) is a major global health concern, particularly due to its association with diabetic kidney disease (DKD). However, the burden of DKD in South Africa remains underreported. This study assessed the prevalence of DKD amongst patients with type 2 diabetes mellitus (T2DM) at Helen Joseph Tertiary Hospital (HJH) and examined its associations with risk factors and target organ damage (TOD).

Methods: This retrospective cross-sectional study reviewed records of 369 patients with T2DM at the HJH Diabetic Clinic from 1 July 2022 to 31 August 2024. DKD was defined as a urinary albumin-creatinine ratio (ACR) ≥ 3 mg/mmol or an estimated glomerular filtration rate (eGFR) < 60mL/min/1.73m^2 on two occasions. TOD was assessed based on documented records.

Results: DKD was identified in 42% of patients (95% CI [37%, 47%]) and was associated with increased age, longer DM duration, and higher systolic blood pressure ($p < 0.001$). No significant differences were observed in HbA1c ($p = 0.24$) and LDL-C levels ($p = 0.18$). Retinopathy ($p < 0.001$), heart failure ($p = 0.018$) and stroke ($p = 0.046$) were more prevalent in the DKD group. Angiotensin-converting enzyme inhibitors or angiotensin receptor blockers were used in 72% and 64% of patients in the DKD and non-DKD groups respectively. Sodium-glucose co-transporter-2 inhibitors were used in 2 patients with DKD and 1 patient without DKD.

Conclusion: DKD is prevalent in patients with DM and is strongly associated with other TOD. Early detection and intervention are essential to improve outcomes and slow disease progression.

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A description of the health profile and associated risk factors in South African university students: a MaRooN health passport study

Category: Clinical

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Background: Non-communicable diseases (NCDs) significantly contribute to premature deaths. NCDs are highly comorbid with common mental disorders (CMDs) and share risk factors. Due to a high prevalence of lifestyle risk factors and CMDs, university students are at risk for developing NCDs. However, this population's NCD prevalence and associated risk factor profile are under-researched. Thus, this study aims to describe the prevalence of NCDs (cardiovascular diseases, chronic respiratory diseases, cancer, and diabetes), symptoms of CMDs, and associated risk factors in university students. It also explores whether latent risk profiles exist based on the observed disease and risk factor variables.

Methods: A cross-sectional descriptive study using 2021–2023 MaRooN Health Passport data was performed using data on the lifetime prevalence of NCDs and CMDs and the prevalence of associated risk factors.

Results: Three thousand two hundred and fifty-two participants (median age = 21 years) completed the survey. Most participants were women (59.7%), undergraduates (76.1%) and attended non-medical faculties (82.2%). The overall lifetime prevalence of NCDs was 36.9%, of which the most prevalent were allergy-related chronic respiratory diseases (CRDs; 34.7%), followed by cardiovascular diseases (2.5%), diabetes (0.8%) and cancer (0.7%). Latent class analysis identified that 40% of participants fell into a higher-risk subgroup, characterised by high levels of psychological distress, poor sleep and symptoms of CMDs. Different risk factor profiles were identified between gender groups.

Conclusion: CRDs are common in this cohort, but the other three groups of NCDs were not. However, these students are at risk of developing them. A latent higher-risk subgroup, characterised by conditions that may be stress-related, can be identified.

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The use of novel biomarkers for assessing glycaemia and diagnosing gestational diabetes mellitus in black African women

Category: Clinical

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Background: This study evaluated the association of IL-6, TNF-α, free fatty acids (FFAs), branched-chain amino acids (BCAAs), and acylcarnitines (C2, C4, C5, and C8-carnitines) with glycaemia during pregnancy, as well as their potential as biomarkers for gestational diabetes mellitus (GDM) in African women.

Methods: This cross-sectional study included two cohorts of women: one in early pregnancy (≤ 20 weeks) and the other in late pregnancy (24–32 weeks). Women in late pregnancy were classified into GDM and non-GDM in accordance with the International Association of Diabetes and Pregnancy Study Groups (IADPSG) criteria. Biomarker levels were compared between GDM and non-GDM groups, and their discriminatory power assessed by receiver-operator characteristic (ROC) curve analysis and their association with glycaemia was measured in both cohorts.

Results: A total of 477 and 170 women in late and early pregnancy, respectively, were included in the analysis. Of the 477 women, 78 (16.4%) had GDM. Women with GDM had significantly greater levels of FFAs and isoleucine, but significantly lower levels of C2, C4, and C5-carnitines than the non-GDM group (all *p*-values < 0.05) but ROC curves demonstrated sensitivities from 7.7% to 44.9% and specificities from 79.3% to 87.7% for GDM diagnosis. In early pregnancy, only valine correlated with blood glucose, independent of BMI and age ($p = 0.029$).

Conclusion: The selected biomarkers are not suitable for diagnosis of GDM. Furthermore, they do not reflect levels of glycaemia in early pregnancy (except for valine) but do so in late pregnancy where they demonstrate altered FFA and BCAA metabolism in those with GDM.

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A complex case of Cushing's syndrome in pregnancy: challenges in diagnosis and management

Category: Clinical

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Introduction: Cushing›s syndrome is a rare endocrine disorder characterized by excessive cortisol production and varied features of protein catabolism. Cushing syndrome is challenging to diagnosed in Pregnancy, as pregnancy is associated with high serum cortisol, non-suppressible dexamethasone test, and mildly elevated UFC and ACTH, posing a challenge in the interpretation of diagnostic tests for Cushing's syndrome, and management in pregnancy is associated with adverse maternal and fetal outcomes.

History and Examination Findings: A 37-year-old gravida 4, para 3 woman presented at 22+4 weeks gestation with clinical features including cutaneous striae, facial plethora, proximal muscle weakness, obesity, hyperandrogenism, and rapid of signs and symptoms of heart failure and new diabetics in pregnancy.

Investigations: A random cortisol (1 144 nmol/L) reference range (138–690 nmol/L), urinary cortisol (1 819 nmol/24h) five times upper limit of normal, and mid night salivary cortisol (79.5 nmol/L) reference range (< 3.6 nmol/L) and ACTH < 1.1 (2–11 pmol/L 8:00 am). Imaging studies revealed a small solid adrenal mass (size 3.6 x 2.8 cm, lesion homogenous, low signal on T2 demonstrates restricted diffusion) on non-enhance MRI and a left ventricular ejection fraction (LVEF) of 45% on echocardiogram. Subsequent decrease in urinary cortisol levels after ketaconazole therapy and on basal bolus insulin and diuretics. Despite challenges, the patient underwent caesarean section at 30 weeks gestation, had fresh stillbirth (FSB). Post-delivery echocardiogram revealed a decline in LVEF to 33%. Post op treatment glycaemic control normal on treat. Follow up contrast CT shows size 2.4 x 1.7 cm, lesion homogenous, low signal and absolute washout < 69%.

Patient currently stable . On anti-failure treatment.

Conclusion: This complex case highlights the challenges of diagnosing and managing CS in pregnancy, particularly in the presence of comorbidities such as heart failure, hyperandrogenism, overt diabetes, and HIV. Early recognition and treatment are crucial to improve maternal and foetal outcomes. Patient awaiting adrenalectomy.

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Chronic kidney disease in diabetic and non-diabetic patients: a tertiary hospital audit

Category: Clinical

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Background: Diabetes mellitus (DM) is one of the leading causes of chronic kidney disease ,contributing to the progression of kidney damage through mechanisms such as hyperglycaemia, hypertension, and altered metabolic pathways. However, CKD

is also prevalent among non-diabetic populations, where various factors contribute to renal dysfunction. This study aims to investigate the differences and predictors of GFR decline in a cohort of participants with and without DM who have chronic kidney disease.

Methods: Retrospective health record review of 196 adult participants with and without DM with a gfr < 60 who were followed up over 5 years. Kidney function and metabolic biomarkers were analysed.

Results: There were 144 participants in non-DM group, 54.8% male. There were 52 participants in DM group 53% male. There were 133 black participants (67.8%). There was a significant difference between mean baseline creatinine in DM vs non-DM of 324 vs 666 respectively ($p < 0.0001$). The mean eGFR in DM vs non-DM group was 23.65 vs 16.24 respectively ($p < 0.0001$). The iPTH was higher in non-DM group ($p < 0.0019$) Vit D levels were significantly lower in DM 45.49 vs 69.85 $p < 0.0001$ FGF23 was lower in DM vs non-DM $p < 0.0025$. There was no difference in haemoglobin, calcium and phosphate between groups.

Conclusion: These results highlight the multifaceted nature of chronic kidney disease and its risk factors and underscores the need for further exploration to understand the mechanisms behind these variations.

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Retinal microvascular complications in children and adolescents with type 1 diabetes mellitus at a quaternary hospital in Pretoria, Gauteng

Category: Clinical

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Background: Diabetes mellitus and its complications are rising among children in sub-Saharan Africa. Diabetic retinopathy (DR), the leading preventable cause of blindness in young adults, requires early detection and management. DR screening implementation remains inconsistent. Identifying risk factors and optimising screening thresholds are crucial for preventing vision loss.

Methods: A retrospective analysis of retinal photographs, demographic data, and clinical profiles of patients under the age of 18 at Steve Biko Academic Hospital's diabetic clinic was performed. We aimed to establish local DR screening guidelines for children and adolescents.

Results: One hundred and one participants with retinal photographs were analysed. Five point nine per cent (5.9%) of participants screened positive for DR, an additional 5% had other eye abnormalities, including cataracts and glaucoma. Sex, tanner staging, cigarette smoke exposure, HBA1C level, serum creatinine, lipogram, and blood pressure were not significant for DR. Older participants ($p = 0.003$), those with longer duration of disease ($p = 0.006$) and those with a lower BMI-for-age ($p = 0.008$) more likely screened positive for DR. The more complications participants had the more frequent DR was present, with 30% of DR positive participants having two or more complications ($p = 0.003$). Microalbuminuria was the only significant predictor for DR after all confounders were factored in (OR = 17.7; 95% CI = 1.01–311.7; $p = 0.049$).

Conclusion: Our findings were consistent with ISPAD guidelines, namely patients with a disease duration of 2 years or more, and age of 12 years or older should undergo screening. Additionally, in our setting, DR screening should be implemented for patients who have microalbuminuria, an increasing number of complications, weight loss or inadequate weight gain.

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Validation of a new phenotypic lipoprotein classification system based on the standard lipid profile in a cohort of patients at a tertiary hospital, South Africa

Category: Clinical

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Background: Cardiovascular disease (CVD) remains a leading cause of morbidity and mortality worldwide. Dyslipidaemia plays a crucial role in CVD pathogenesis, necessitating effective diagnostic and management strategies. Recently Sampson et al. proposed a new phenotypic lipoprotein classification system based on standard lipid profiles to improve diagnostic accuracy when evaluating patients with dyslipidaemias. The aim of the study was to validate this new classification system, using standard lipid profiles assessing its correlation with gel electrophoresis in diagnosing dyslipidaemias.

Methods: This was a retrospective observational study conducted in an academic tertiary laboratory, South Africa. Two datasets were analysed; 650 standard lipid profiles and 100 paired lipid profiles with serum lipoprotein gel electrophoresis results. Phenotypes were classified based on triglyceride and non-HDL cholesterol levels using cut-offs previously derived by Sampson et al. Chi-square tests and Cohen's Kappa were used to evaluate agreement between the lipid profile-based classification system and gel electrophoresis.

Results: The new classification identified 72% of subjects as normolipidemic, with Type IVb (15.7%) as the most prevalent abnormal phenotype. Significant differences were observed between the two methods ($\chi^2 = 85.80$, $p < 0.000001$). Type III phenotype was excluded due to overlapping values with other categories.

Conclusion: The new phenotypic system offers a practical alternative for resource-limited settings, highlighting the value

of triglycerides and non-HDL-C in evaluating patients with dyslipidaemia. Observed discrepancies with gel electrophoresis were attributed to limited data points that were available at the study site, thus further validation studies are needed to ensure comprehensive risk stratification and clinical applicability.

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Laboratory screening of porphyrias using HPLC with a fluorescence detector: a case of acute intermittent porphyria in a 17-year-old female

Category: Clinical

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Introduction: Porphyrias are a group of inherited disorders of heme biosynthesis. They are caused by a deficiency of specific enzymes in the heme pathway that lead to overproduction of intermediates or precursors (porphyrins, delta-aminolevulinic acid [ALA], porphobilinogen [PBG]), which accumulate in body organs (liver, red blood cells). Porphyrias are classified according to their clinical manifestations as neuro-visceral (acute attacks), cutaneous (photosensitivity) and mixed types. There are various methods for the detection of porphyrins in body fluids. Our laboratory uses high-pressure liquid chromatography (HPLC) for detection of porphyrin precursors in urine samples, after a positive PBG screening using the Hoesch test.

History and examination findings: A 17-year-old female presented at Baragwanath Academic Hospital with seizures, acute severe abdominal pain, nausea, vomiting, tachycardia and increased blood pressure. The urine tested positive for PBG using the Hoesch test and was further analysed by HPLC for fractionated porphyrins using a fluorescence detector.

Investigations: The fractionated HPLC showed a significant increase in uroporphyrins, increased copro-porphyrins and a mild increase in pentacarboxyl porphyrins. These findings, together with the patient's clinical history, were highly suggestive of acute intermittent porphyria (AIP).

Conclusion: Fractionated HPLC, using a variety of detectors, is an efficient method for testing of porphyrias. With the use of a fluorescence detector, our laboratory was able to detect a significant increase in uroporphyrin, coproporphyrin and pentaporphyrin fractions, which are strongly suggestive of the diagnosis of AIP. Appropriate treatment of the patient was instituted which resulted in resolution of symptoms. Genetic testing can assist in confirming the diagnosis of AIP.

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Functional gonadotroph adenoma: an interesting presentation of a follicle stimulating hormone (FSH) secreting tumour

Category: Clinical

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Background: Functional gonadotroph adenomas (FGA) are rare benign tumours of the pituitary. Unlike other functional pituitary adenomas that present with characteristic symptoms related to excess hormone secretion, these tumours are usually clinically silent. Thus, patients often present late with pressure symptoms, similarly to non-functioning adenomas. Clinical syndromes due to hypersecretion of intact gonadotropins are rare.

Methods: A 41-year-old man presented at the Endocrinology Clinic of a tertiary hospital complaining of blindness of the left eye, and deteriorating vision on the right. No symptoms of hypopituitarism, or impotence were elicited from the history. He had normal vitals (BP = 135/82, Pulse = 82) with no features of Cushing's disease, acromegaly, hypogonadism nor hypothyroidism. Fundoscopic evaluation revealed optic atrophy with complete blindness of the left eye, and temporal outfall of the right observed on formal visual field testing.

Results: An MRI scan showed a hyperdense sellar mass with suprasellar extension measuring 41 X 49 X 49 mm, with peripheral calcifications. Biochemical evaluation he had elevated follicular stimulating hormone (FSH) of 18.1 IU/L (Reference interval [RI] 1.0–12.0), a normal luteinising hormone (LH) of 1.4 IU/L (RI 0.6–12.1), total testosterone level of 11.6 nmol/L (RI 11.6 (RI 8.8–27.9) and sex hormone binding globulin of 26.1 nmol/L (RI 16.2–68.5). The normal LH and testosterone levels excluded primary hypogonadism, and the discordancy between the high FSH and low-normal LH levels were suggestive on an FSH secreting gonadotroph adenoma. The patient's kidney function and the rest of the pituitary hormonal evaluation were within normal limits. Ultrasound evaluations were indicative of testicular enlargement especially on the right (right testis 30.31 ml, left 18.70 [normal volume 12.5–19 ml]). Histological evaluation of the sample obtained during surgery was consistent with a pituitary adenoma and immunohistochemical staining positive for FSH. Post resection, FSH levels decreased to 8.3 IU/L [RI 1.0–12.0], though residual tumour was visualised on imaging.

Conclusion: Biochemical features and evidence of testicular enlargement were consistent with FGA. Given the risk of recurrence of these tumours, long-term imaging and biochemical investigations are required.

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Pituitary stalk interruption syndrome in adulthood

Category: Clinical

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Background: Pituitary stalk interruption syndrome (PSIS) is a rare congenital disorder characterised by the radiological triad of an absent or thin pituitary stalk, ectopic or absent neurohypophysis, and hypoplastic or absent adenohypophysis. This anatomical anomaly leads to varying degrees of pituitary hormone deficiencies, resulting in growth restriction, delayed puberty, and other endocrine dysfunctions. This report describes a patient with PSIS, missed in childhood, and later diagnosed in adulthood.

History and examination: A 25-year-old lady was referred the endocrinology clinic with a history of delayed growth and primary amenorrhoea. On examination, she had proportionate short stature and Tanner stage 1 breast and pubic hair development.

Investigations: Laboratory tests were consistent with secondary hypothyroidism (TSH mIU/L; free T4 9.3 pmol/L), growth hormone deficiency (insulin-like growth factor-1 < 20 mcg/L) and hypogonadotropic hypogonadism (follicle stimulating hormone 2.3 IU/L, Luteinizing Hormone 0.8 IU/L, oestradiol pmol/L). Prolactin (11.3 mcg/L) and 9 am serum cortisol (176 nmol/L) were normal. Glucagon Stimulation Test confirmed growth hormone deficiency (peak growth hormone < 0.1 mcg/L). 1 mcg synacthen test showed an adequate cortisol response (764 nmol/L at 30 minutes). Pituitary MRI showed an absent pituitary stalk, ectopic neurohypophysis at the medial eminence/floor of the third ventricle and hypoplastic adenohypophysis.

Conclusion: PSIS is a rare cause of multiple pituitary hormone deficiencies, with fewer than 1 000 reported cases. Presentation may occur in early infancy (with hypoglycaemia, prolonged jaundice, microphallus and cryptorchidism) or in childhood/adolescence (with short stature and delayed puberty). Early clinical diagnosis is crucial to initiate timeous hormonal replacement, to optimize growth and sexual development.

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Phaeochromocytoma and Tetralogy of Fallot: two case reports

Category: Clinical

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Introduction: Phaeochromocytoma and paraganglioma are rare neuroendocrine tumours that originate from neuroectodermal chromaffin cells of the medulla. Tetralogy of Fallot (TOF) is a cyanotic congenital heart disease which consists of four abnormalities: pulmonary outflow tract obstruction, ventricular septal defect, overriding aortic root and right ventricular hypertrophy. Although the occurrence of phaeochromocytoma and TOF concurrently has been reported, this is a rare occurrence. We report two cases with this and discuss possible pathophysiology. It is speculated that chronic hypoxia gives rise to adrenal medullary hyperplasia and eventually development of phaeochromocytoma. However, the detailed mechanism of congenital heart disease leading to phaeochromocytoma has yet to be explained.

History and examination findings: Case 1: A 48-year-old female known with an unrepaired Tetralogy of Fallot complicated by heart failure with baseline NYHA 2 and saturation on room air around 80%. The cyanotic heart disease is complicated by polycythemia with no hyperviscosity symptoms. The patient presented with episodes of intermittent palpitations and mild headaches. Case 2: A 48-year-old female known with Tetralogy of Fallot which was repaired at the age of 5 years. The patient had renal impairment and incidental adrenal masses were noted on during for investigation of the renal impairment. Additionally, the patient has hypertension, type 2 diabetes mellitus, as well as episodes of ventricular tachycardia and chronic atrial fibrillation on lifelong warfarin. The patient reported symptoms of palpitations with associated diaphoresis and vomiting. During admission to hospital the patient had episodes of tachycardia and elevated blood pressure especially nocturnal.

Investigations: Case 1: Results of 24-hour urine catecholamines confirmed excess normetanephrines and metanephrines (table). An adrenal protocol CT showed bilateral adrenal masses with and delayed wash out in keeping with pheochromocytoma. A 68-Ga-DOTANOC PET-CT demonstrated uptake in the known bilateral adrenal masses as well as intense further uptake in bilateral paravertebral masses (four in total), in the carotid space suggestive of paragangliomas and in the liver suggestive of metastatic disease. Case 2: Serum and urine catecholamine levels were elevated (table). An adrenal protocol CT demonstrated a left supra renal mass with a size of 2.2 cm x 2.6 cm x 3.7 cm and density consistent with phaeochromocytoma. A 68-Ga-DOTANOC PET-CT demonstrated intense uptake on the left adrenal gland.

Conclusion: We report two cases of confirmed phaeochromocytoma in patients with Tetralogy of Fallot, one whose defect was repaired in childhood, though with complications, and one never repaired. This is to our knowledge the first report of cases in sub-Saharan Africa.

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Perceptions and experiences of Continuous Glucose Monitoring for diabetes management in the public healthcare sector in South Africa: a patient and healthcare provider perspective

Category: Clinical

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Background: Despite the benefits of Continuous Glucose Monitoring (CGM) in diabetes management, its adoption in low- to middle-income countries remains low. This study explored the perceptions and experiences of CGM use among people living with diabetes (PLWD) and healthcare providers (HCPs) in a three-arm pragmatic randomized controlled trial in South Africa.

Methods: We conducted 15 focus groups discussions (FGDs) and 16 semi-structured interviews (SSIs) across three study sites. Data analysis followed thematic content analysis. The findings are presented according to the socioecological framework.

Results: A total of 109 participants including 76 PLWD, 17 caregivers and 16 HCPs were included. CGM users highlighted the convenience of CGMs, which minimised frequent fingerpricks and simplified glucose monitoring. However, individual factors such as unemployment and food insecurity posed significant barriers to optimal CGM use. Personal motivation was critical, particularly among adolescents for self-management using CGM. Interpersonal barriers included lack of social support and perceived stigma. Continuous CGM training and diabetes education, as well as positive relationships with HCPs were motivating factors at the health system level. HCPs emphasized the need for a multidisciplinary care team, including diabetes nurse educators. At the policy level, cost was perceived as a significant barrier to access in the public health sector.

Conclusion: Our findings demonstrate that the introduction of CGMs in the public health sector in South Africa is feasible and acceptable. However, a multi-level approach is critical to addressing the complex, interrelated factors that may influence the successful integration of CGMs into diabetes management in the public health sector.

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The prevalence of hypothyroidism in patients with type 2 diabetes mellitus attending the diabetic clinic at Helen Joseph Tertiary Hospital

Category: Clinical

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Background: The purpose of this study was to determine the prevalence of hypothyroidism in patients with Type 2 Diabetes Mellitus (T2DM) attending the diabetic clinic at Helen Joseph Tertiary Hospital.

Methods: This was a retrospective clinical audit for the period 01 January 2020 to 30 April 2020. Three hundred consecutive patients were screened, and study participants were restricted to those aged 45 years or older with T2DM. Those who were pregnant or acutely ill were excluded. The SEMSDA-ACE/SA guidelines for normal TSH values were used to qualify hypothyroidism. Continuous variables were compared between groups using Wilcoxon rank sum tests with continuity correction.

Results: Of the 290 participants who met the inclusion criteria the prevalence of hypothyroidism was 22.8%. Of these 66 participants, 20 (30.3%) were known with hypothyroidism and 46 (69.7%) were newly diagnosed based on TSH targets. The prevalence of hypothyroidism was 28.9% among participants younger than 65 years and 11.7% among participants aged 65 years and older. The median (IQR) HbA1c of those with (9.4% [8.1–10.8%]) and without hypothyroidism (9.3% [7.8–11.1%]) were similar (p = 0.838). Only 19 (28.8%) participants with hypothyroidism were on treatment. Of these only 4 (21.1%) had TSH values within their age appropriate target range, with 8 (42.1%) having TSH levels above the target range.

Conclusion: The prevalence of hypothyroidism in this study was found to be higher than in similar international studies. The low treatment rates and suboptimal levels of control of hypothyroidism highlights the need for clinicians to be more vigilant in screening and correctly managing this population group.

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An overview of hypothyroid disorders in the paediatric endocrine unit at Chris Hani Baragwanath Academic Hospital between 2011-2023

Category: Clinical

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Background: Hypothyroidism is a prevalent endocrine disorder in the paediatric population, but data on the spectrum of this disease in the general paediatric population in South Africa is limited.

Objective: To describe the demographics, types, clinical features, co-morbidities and outcomes of paediatric hypothyroidism at Chris Hani Baragwanath Academic Hospital (CHBAH).

Methods: A retrospective, descriptive study was conducted at CHBAH's Paediatric Endocrine Unit. Of 246 identified files from 1 January 2011 to 31 December 2023, 147 met inclusion criteria and were categorised into subclinical, central, primary and acquired hypothyroidism groups; statistical significance set at $p < 0.05$.

Results: Of 147 patients, 76 were male (51%). Subclinical hypothyroidism (SH) was most common (45%), followed by central (24%), primary (20%), and acquired hypothyroidism (11%). Median age at diagnosis was 1.17 years (IQR 0.34–3.63). Neuromuscular symptoms were the most common clinical presentation (58%). Down syndrome was the most common

co-morbidity (48%) and strongly associated with SH ($p < 0.001$). On analysis of all patients, levothyroxine was initiated in 74%, with a mean starting dose of 4.39 µg/kg/day (± 2.97) — lower than guideline recommendations. Self-reported treatment compliance was 63%, with biochemical adherence in 87% of these. Growth velocity declined slower in treated vs untreated patients ($p < 0.001$). Neurodevelopmental improvement was reported in 51% of treated patients, mainly in motor and communication domains.

Conclusion: SH was the predominant form of hypothyroidism, with Down syndrome as the most common co-morbidity. Levothyroxine improved neurodevelopment and growth. Findings support the need for formal neonatal screening for early detection and treatment of congenital hypothyroidism.

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Hypothyroidism: an unusual cause of massive pericardial effusion in the South African setting

Category: Clinical

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Background: A 40 year old female presented with features of heart failure, with echocardiogram revealing a large pericardial effusion (4,4 cm anteriorly; 3.1 cm over the apex; 5,8 cm posteriorly). There were no clinical features suggestive of tamponade. Workup excluded commoner aetiological causes and the diagnosis was narrowed down to that of severe hypothyroidism.

Methods: The patient presented with 1 week history of orthopnoea, exertional dyspnoea, facial and lower limb swelling, on a background of chronic fatigue, constipation and weight gain. Clinically she had signs of heart failure: peripheral oedema, basal crepitations and clear heart sounds with a raised JVP; along with signs of hypothyroidism: coarse facial features, dry skin, peripheral and peri-orbital oedema, proximal weakness and delayed relaxion of deep tendon reflexes.

Results: The patient›s TSH levels were severely elevated at > 48.10 mIU/L (normal range 0.34–5.6 mIU/L) with a T4 of 1.5 pmol/L (normal range 7.6–16.1 pmol/L), confirming severe hypothyroidism. Commoner causes of pericardial effusion were eliminated with a negative RVD test, normal urea levels, low CRP levels, negative TB testing on both sputum and pericardial fluid, negative auto-immune studies, and normal troponin levels. A pericardiocentesis was performed, and drained a golden exudative fluid amounting to 1 250 ml over 3 days.

Conclusion: Following pericardiocentesis and initiation of thyroid hormone treatment (100 ug of levothyroxine daily), there was clinical improvement in the patient›s condition. Thyroid antibodies were investigated and were pending as of discharge. Hypothyroidism should be considered as a diagnosis of exclusion in patients presenting with pericardial effusion. Thyroid hormone replacement is essential. Regular monitoring and follow-up are crucial to ensure therapeutic efficacy and to prevent potential complications such as cardiac tamponade.

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Evaluating the prevalence of obstructive sleep apnoea in people living with type 2 diabetes at Helen Joseph Hospital, Johannesburg, South Africa

Category: Clinical

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Background: Obstructive sleep apnoea (OSA) and Type 2 diabetes (T2D) are interrelated conditions, with T2D increasing the risk of OSA through weight gain and autonomic dysfunction, while OSA is diabetogenic through dysregulated sleep. It is estimated that nearly 50% of T2D have OSA. This study evaluated the prevalence of OSA amongst patients at the diabetic clinic, Helen Joseph Tertiary Hospital.

Methods: A total of 349 patients with T2D were screened using the STOP-BANG score. Data on blood pressure, body mass index (BMI), comorbidities, and social habits were collected, along with laboratory results for HbA1c, LDLand TSH levels.

Results: Findings revealed 171 patients (49%) tested positive for OSA. Common comorbidities included hypertension (40.9%) and hypothyroidism (28.1%). Patients with OSA engaged in 1.09 fewer hours of weekly exercise ($p = 0.0047$). There were no significant differences in Hba1c, average blood pressure, neck circumference, or BMI between patients with and without OSA.

Conclusion: This study corroborates global trends of high OSA prevalence in patients with T2D. As no association between BMI and neck circumference were found in this study (also noted in international data), it suggests other factors may contribute to OSA in this population. The significant prevalence emphasises the necessity for enhanced OSA screening in diabetic clinics.

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Diagnosis of Cushing's syndrome in chronic kidney disease

Category: Clinical

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Background: Chronic kidney disease (CKD) is associated with non-neoplastic hypercortisolism due to activation of the hypothalamic-pituitary- adrenal axis, and confounds the interpretation of diagnostic tests for hypercortisolism. A

diagnostic challenge of Cushing's disease in a patient with CKD is presented.

Clinical history: A 55-year-old woman with Type 2 diabetes, HIV (on a protease-inhibitor) and CKD stage 4, presented with fatigue, limb weakness, polyuria and polydipsia. Her phenotype was lipodystrophic with facial lipoatrophy, a dorsocervical fat pad, central obesity and thin limbs, but lacked catabolic signs (skin thinning, striae or bruising). Elevated 9am cortisol (948 nmol/L) and inappropriate ACTH (39.7 pmol/L) necessitated further investigation.

Results of investigations: Screening tests confirmed hypercortisolism (serum cortisol 763 nmol/L following the 1 mg overnight dexamethasone suppression test; elevated midnight serum cortisol of 706 nmol/L). Discrepant 24 hour urine free cortisol results were attributed to CKD (253 nmol/24 hours and 64 nmol/24 hours). Midnight salivary cortisol was rejected. Rise in ACTH > 6 pmol/L during the desmopressin test suggested neoplastic rather than non-neoplastic hypercortisolism; due to a eutopic source (63% rise in ACTH and 30% rise in cortisol at 30 minutes). Non-contrast MRI confirmed a pituitary macroadenoma. Medical management was instituted with cabergoline and the patient was referred for neurosurgical evaluation.

Conclusion: Chronic inflammation drives a state of non-neoplastic hypercortisolism in CKD. Diagnosis is confounded by reduced GFR, which interferes with all modalities of cortisol assessment. Late night salivary cortisol and the 1 mg overnight dexamethasone are the recommended screening tests in CKD, but more studies are required to establish disease-specific cut-offs.

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Prevalence of symptomatic polyneuropathy in patients with type 2 diabetes mellitus attending the diabetic clinic at Helen Joseph Tertiary Hospital, South Africa

Category: Clinical

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Background: Diabetic neuropathy has an estimated prevalence of 50% amongst longstanding people living with diabetes (PLWD); with distal symmetric polyneuropathy (DSPN) being the most common manifestation. Poor glycaemic control is a known risk factor for DSPN development. This study aimed to determine the prevalence of symptomatic DSPN in patients with type 2 diabetes mellitus (T2DM) using a validated symptom screening questionnaire – the Diabetic Neuropathy Symptom (DNS) score. Additionally, the association between Haemoglobin A1c (HbA1c) and DSPN was investigated.

Methods: A cross-sectional study was performed at the diabetic clinic at Helen Joseph Tertiary Hospital, Johannesburg, South Africa. Two hundred and six consecutive patients with T2DM were included in the study. Underlying comorbidities and HbA1c values were obtained from patient records. The DNS score was used to assess for the presence of symptomatic DSPN. The prevalence of DSPN was determined as the number of patients who had a positive DNS score.

Results: The prevalence of symptomatic DSPN in the study population was 61.2%. Amongst those who screened positive for DSPN, 58% were not receiving pharmacological treatment. Patients with HbA1c values between 7–10% and > 10% were 2.9 and 3.7 times (respectively) likely to have DSPN (PR =2.9, $p = 0.001$; PR 3,7 $p = < 0.001$, respectively), compared to those with an HbA1c < 7%.

Conclusion: A higher than expected prevalence of symptomatic DSPN was observed in this study population, indicating the need for enhanced screening efforts. A significant proportion of symptomatic patients were not receiving treatment. Poor glycaemic control with HbA1c values > 7% significantly increases the risk of DSPN.

https://doi.org/10.36303/JEMDSA.2025.30.1.P31

Evaluation of the relationship between diabetes control and oral health in the Kalafong Provincial Tertiary Hospital Diabetic Clinics

Category: Clinical

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Background: Gum disease and periodontitis are major contributors to morbidity in patients with diabetes mellitus (DM). The association between DM and periodontitis is known to be bidirectional. Gum disease and periodontitis lead to poorer glucose control in patients with DM and also cause insulin resistance in those without DM. Uncontrolled DM is both a risk factor and a modifying factor for gum disease and periodontitis when present, it leads to more severe forms of the disease.

The relationship between DM and oral health in adult patients with DM type 1 and 2 and uncertain aetiology attending the Kalafong Provincial Tertiary Hospital (KPTH) has never been explored.

Methods: This is a cross-sectional study conducted in KPTH in the Internal Medicine diabetes clinics. The participants were adult patients with type 1, type 2 DM, and DM of unknown aetiology.

Results: Five hundred and twenty-six patients with DM were included. The mean HbA1c in these patients is 9.3. Glycated haemoglobin (HbA1c) did not differ significantly between participants with gum disease and periodontitis and those without. No significant difference was observed in the HbA1c levels between patients with (HbA1c 9.5%) and without (9.35%) any form of gum disease ($p = 0.086$), or between patients with (HbA1c 9.47%) and without (9.12%) any loose teeth ($p = 0.906$).

The difference in HbA1c between patients with (HbA1c 9.3%) and without (HbA1c 9.35%) missing teeth was statistically significant ($p = 0.025$), indicating patients with missing teeth had a slightly lower HbA1c.

Conclusion: The findings showed that, no difference in glucose control could be demonstrated between patients with and without poor oral and dental health.

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A rare case of hypothyroidism-induced cardiomyopathy at a tertiary institution in South Africa

Category: Clinical

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Introduction: Dilated cardiomyopathy is a frequently diagnosed medical condition in hospitalised patients and is associated with a broad list of aetiologies. While most causes are irreversible, evaluation of reversible aetiologies such as thyroid disease is essential to improve the prognosis of these patients. Although severe thyrotoxicosis may often present with heart failure, hypothyroidism as a cause of dilated cardiomyopathy is rare and limited to a few case reports. This case report highlights hypothyroidism as a potential cause of cardiomyopathy as well as the impact of appropriate treatment on the improvement of cardiac function.

History and examination findings: This is a case report of a 30-year-old male, with no significant cardiovascular risk factors, who presented with a two-month history of progressive dyspnoea and anasarca, on further enquiry he reported cold intolerence, constipation and dry skin. Clinical examination was in keeping with biventricular failure due to a dilated cardiomyopathy.

Investigations: Biochemical testing confirmed overt hypothyroidism (TSH > 100 mIU/L [normal: 0.27–4.2], fT4 < 1 pmol/L [normal: 11.9–21.6]). The echocardiogram was compatible with a dilated cardiomyopathy: four chamber enlargement with global hypokinesia and an ejection fraction of 23%. Intravenous diuretics and oral levothyroxine were commenced. However, there was a diminished response to diuretics that resolved only once fT4 levels were in the normal range. On anti-remodelling therapy and levothyroxine replacement, cardiac failure resolved. The ejection fraction improved to 48% after 6 months.

Conclusion: Severe overt hypothyroidism may present as a reversible dilated cardiomyopathy. Response to diuretics may be attenuated until thyroid hormone is in the normal range.

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An audit of the fracture liaison service at Helen Joseph Tertiary Hospital in Johannesburg, South Africa

Category: Clinical

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Background: Fragility fractures, especially in older adults, lead to significant morbidity, with 75% of patients experiencing complications within a year. Fracture Liaison Services (FLS) are considered best practice for managing these fractures, focusing on identifying osteoporosis causes, initiating early treatment, and assessing fall risks. While FLS is standard in many developed countries, there is limited data on these services in Africa, particularly South Africa.

Methods: This retrospective audit reviewed the newly implemented FLS at Helen Joseph Hospital (HJH) from October 2022 to March 2024. It included post-menopausal women and men over age 50 admitted with fragility fractures.

Results: A total of 247 patients were analysed, 74% of whom were female. Primary risk factors included insufficient physical activity (53.1%), smoking (52.9%), previous fractures (25%), and falls (42%), with 48% of males reporting significant alcohol use. The hip was the most common fracture site (59.9%). Vitamin D deficiency (< 50 ng/ml) was documented in 41.1% of patients, with no ethnic differences. The average Fracture Risk Assessment Tool score was 11.3, a threshold of 20 is typically used to initiate treatment in patients within the intermediate risk category. Only 27.8% received zoledronic acid before discharge, largely due to medication shortages (72.6%) and renal dysfunction (27.4%). In-hospital mortality was 5%.

Conclusion: The FLS at HJH reveals significant challenges in managing fragility fractures in the public sector, including medication shortages and high prevalence of Vitamin D deficiency. The study underscores the need for improved strategies in South Africa, providing crucial insights for future service development and research.

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Oedema in an adolescent with type 1 diabetes mellitus: a case report from South Africa

Category: Clinical

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Introduction: The introduction of insulin significantly reduced the mortality and morbidity associated with type 1 diabetes mellitus. However, it does have adverse effects such as hypoglycaemia and weight gain. Insulin oedema is a rare but important complication of insulin therapy. The pathophysiology remains unclear, but many hypotheses have been proposed.

History and examination findings: We report the case of a 15-year-old male with poorly controlled type 1 diabetes mellitus who presented with swelling and shortness of breath. Clinical examination revealed extensive pedal oedema extending to the sacral area, along with tender hepatomegaly and abdominal swelling.

Investigations: The workup for other potential causes of oedema yielded negative results. Cardiac and renal evaluations were unremarkable. However, the liver enzymes were moderately elevated with no clinical or biochemical features of liver failure present. Imaging revealed a congested liver with no significant structural abnormalities. A full hepatitis panel was done, and infectious and autoimmune causes were ruled out. Conclusion: He was managed conservatively with fluid and salt restriction. Both the transaminitis and the oedema resolved within 21 days. Insulin oedema is rare and presents a challenging diagnosis, as its pathophysiology is poorly understood. It is important to rule out other potential causes of oedema before reaching this diagnosis. The associated transaminitis in this case added to the diagnostic complexity. This case underscores the need for clinicians to be aware of insulin oedema as a potential complication in patients on insulin therapy. Conservative management with fluid and salt restriction may be sufficient and is often the only treatment required.

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The incidence of ECG changes suggestive of myocardial infarction in patients attending Kalafong Provincial Tertiary Hospital Diabetes Clinic

Category: Clinical

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Background: Diabetes mellitus (DM) is a risk factor for myocardial infarction (MI). This study focuses on the electrocardiographic (ECG) changes suggestive of MI, as well as the effects of DM on the heart.

Methods: Patients aged 30 years and older were selected from the DM clinic database covering the period from 2008 to 2023. A total of 634 out of 732 were included in the study. ECG abnormalities were reviewed and discussed with the senior physician, with findings documented in the electronic database. Two ECGs, at least one year apart, were compared. Kaplan-Meier analyses were performed to assess the time to new ischemia and MI in patients without baseline ECG abnormalities comparing patients with Type 1 and Type 2 DM.

Results: ECGs at baseline were normal in 83% (n = 568) of the patients. Among them, 33.1% (n = 210) developed ECG abnormalities at follow-up. Ninety-seven patients (15.3%) showed ECG signs suggestive of MI, with 66 (16.7%) females and 31 (13%) males exhibiting these changes. The most common myocardial regions affected were the inferior wall, followed by the infero-lateral wall, while the anterior wall was the least commonly affected. Type 2 DM patients exhibited a higher incidence of ECG changes suggestive of MI compared to those with Type 1 DM (16.7% vs. 9.8%). Other common ECG abnormalities included the right and left bundle branch blocks; first-degree AV block; P mitrale; increased incidence of left ventricular hypertrophy (LVH); and poor R wave progression.

Conclusion: A higher incidence of MI and other ECG abnormalities were observed in Type 2 DM.