



*Abstracts from the*

**Student Medical Summit 2025**

**Date:** Saturday, February 15<sup>th</sup> 2025

**Location:** UCD Campus in Belfield, Dublin 4, Ireland.

## **Evaluating treatment approaches for adults with inherited factor X deficiency: a review of current literature**

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### *Aims*

Inherited Factor X Deficiency (FXD), an autosomal recessive coagulation disorder due to mutations in F10 gene. FXD has a worldwide prevalence of 1:1,000,000, higher in countries with higher rates of consanguineous marriage<sup>1</sup>. Classification of severity depends on baseline Factor X, patients with <10% experience high risks of spontaneous bleeds and require regular prophylaxis. Life-threatening consequences such as intracerebral bleeding can occur early in life, followed later in life by haematomas, menorrhagia and spontaneous hemarthroses severely affecting quality of life<sup>2</sup>.

### *Methods*

This literature review appraises current treatment approaches. Search strategy was employed (Pubmed, Cochrane Library, Clinicaltrials.gov) using keywords “FXD”, “single factor X concentrate”, “quality of life” and “treatment”. Literature screened using inclusion/exclusion criteria, focusing on women with FXD and treatment efficacies, summarised in Figure 1.

### *Results*

Single factor X concentrate, the most common novel FDA-approved treatment in Ireland may provide safe, selective less-demanding bleed protection in adults especially in women with menorrhagia and pregnancy-related complications. Other alternatives (fresh frozen plasma, multiple factor concentrates) used depending on patient co-morbidities that affect bleeding risk<sup>3</sup>.

### Discussion

Many safe alternatives can be used but require personalisation and dosage changes. Further studies required to analyse long-term effects of FXD treatment, adverse events and patient needs to decrease the burden of disease that FXD causes with the risk of life-altering consequences.

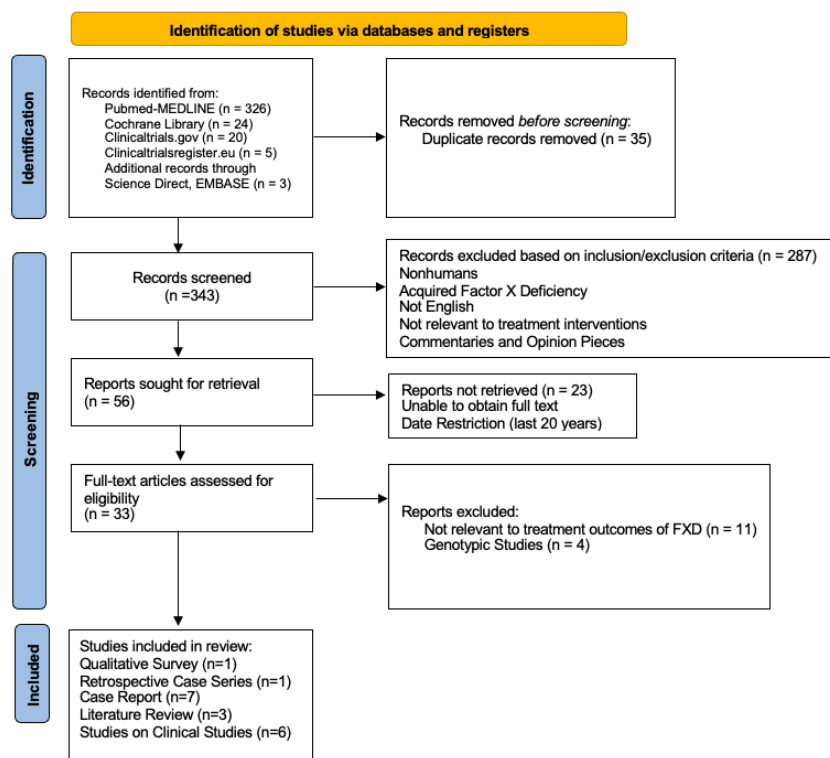


Figure 1: Prisma Flow Diagram

### References:

1. Mumford AD, Ackroyd S, Alikhan R, Bowles L, Chowdary P, Grainger J, et al. Guideline for the diagnosis and management of the rare coagulation disorders. British Journal of Haematology. 2014 Aug 6;167(3):304–26.
2. Peyvandi F, Di Michele D, Bolton-Maggs Phb, Lee Ca, Tripodi A, Srivastava A. Classification of rare bleeding disorders (RBDs) based on the association between

coagulant factor activity and clinical bleeding severity. *Journal of Thrombosis and Haemostasis*. 2012 Sep;10(9):1938–43.

3. O’Connell N, Adults with Haemophilia and Related Bleeding Disorders Acute Treatment Guidelines. National Haemophilia Council Guidelines 2023 March 14;7.0:46-50

## **Assessing medication adherence and beliefs about medicines in patients with Primary Biliary Cholangitis being treated with Ursodeoxycholic Acid monotherapy**

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### **Abstract**

#### *Introduction*

Ursodeoxycholic acid (UDCA) is the first line treatment for PBC, however up to 40% of patients have insufficient response. Obeticholic acid, reimbursable since 2022, is a costly second-line option. As poor adherence negatively impacts response rates, understanding compliance with UDCA is crucial to inform prescribing decisions for obeticholic acid.

#### *Aim*

To use the Beliefs about Medicines Questionnaire (BMQ) and the Medication Adherence Rating Scale (MARS-5) to identify rates of low adherence, characterise risk factors, and identify barriers to adherence.

#### *Methods*

This was a cross-sectional study at a tertiary hospital involving PBC patients on UDCA monotherapy. Questionnaires were mailed with return envelopes. Descriptive statistics

summarised demographics and Cronbach alpha was calculated. A MARS-5 total (5-25) was calculated, and the median score was used to define 'high' adherence. Pearson's correlation assessed BMQ and MARS-5 subscales, and binary logistic regression analysed associations between high adherence, demographics, and BMQ scores

### *Results*

Response rate was 41% and Cronbach alpha was acceptable ( $>0.65$ ). The median score of the MARS- 5 (24) defined 'high' adherence, observed in 72.6% of respondents. BMQ-General benefit score correlated positively with the MARS-5 score (Pearsons  $=0.3$ ,  $p$ -value  $=0.02$ ). Logistic regression shows a statistically significant relationship between General-Benefit score and high adherence (OR = 1.81,  $p$ -value = 0.012).

## **Endovascular management of non-cirrhotic acute portomesenteric venous thrombosis**

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### **Abstract**

#### *Aims*

Discuss the pathophysiology of acute PMT in non-cirrhotic patients and address endovascular treatment options. Delve into the significance of acute PMT in the context of COVID-19 infection, vaccine-induced immune thrombotic thrombocytopenia, and liver transplantation.

#### *Background*

Acute portomesenteric venous thrombosis (PMT) is the partial or complete occlusion of the portal veins or their tributaries. It is a distinct entity in non-cirrhotic versus cirrhotic patients.

The distinguishing pathophysiologic feature leading to acute PMT in cirrhotic patients is venous flow stasis secondary to portal hypertension. It is potentially fatal as it can result in bowel ischemia. Anticoagulation is the first-line treatment; however, there are no guidelines for patients with contraindications or who do not improve. The heterogeneous etiologies in non-cirrhotic patients have precluded randomized controlled trials to evaluate optimal interventional treatment. Nonetheless, recent evidence indicates that intervention yields favorable outcomes.

### *Results*

The authors propose a treatment algorithm, adapted from the Baveno VII treatment recommendations, to guide clinicians. A distinguishing aspect of treatment in these patients is determining factors indicating if patients are presenting with reversible bowel ischemia vs. irreversible necrosis to guide clinical decisions regarding surgical intervention necessity.

### *Discussion*

CDL and TIPS creation are mainstay interventions for acute PMT. Antegrade and retrograde access approaches represent additional strategies for recanalization. Emerging techniques such as large-bore thrombectomy show promise in small-scale studies. These technique applications are crucial in specific clinical scenarios.

## **Tele-emergency Medicine: a systematic review of the impact of telemedicine on emergency medicine on quality of care, time to treatment, and accessibility versus traditional care**

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### **Abstract**

#### *Background*

Telemedicine has surged in popularity since the COVID-19 pandemic. In Emergency Medicine (EM), telemedicine—referred to as ‘tele-EM’—enhances patient flow and potentially reduces overcrowding by enabling timely remote consultations. This study systematically reviews the impact of tele-EM, focusing on quality of care, time to treatment, and accessibility compared to traditional in-person care.

#### *Methods*

Following PRISMA guidelines, a systematic review was conducted, searching four electronic databases: PubMed, Scopus, CENTRAL, and Embase. Search terms included ‘telemedicine’, ‘telehealth’, ‘tele-emergency’, ‘emergency departments’, ‘quality of care’, ‘implementation’, and ‘impact’. Studies focused on tele-EM interventions were included. Exclusion criteria

included non-EM populations and failure to meet primary endpoints. Cohort studies were assessed for bias using the Newcastle-Ottawa Scale. (Figure 1)

### Results

Of the 1,195 studies identified, 17 met the inclusion criteria. (Figure 2) Tele-EM showed significant benefits, including a 30% decrease in paediatric interfacility transfers. Tele-EM shortened the time to treatment, with a 20-minute reduction in time-to-ECG for myocardial infarction patients and a 50% reduction in time-to-head CT interpretation for neurological emergencies. Tele-EM also enhanced clinical decision-making by 20% for chest pain patients. Real-time video conferencing was the most utilized modality.

### Discussion

Tele-EM demonstrates significant potential in EM. It improves accessibility, reduces wait times, and enhances patient outcomes while potentially alleviating emergency department (ED) overcrowding through admission avoidance. Future research should prioritize high-quality randomized trials to support broader implementation of tele-EM across diverse healthcare settings.

Study	Selection (0-4)	Comparability (0-2)	Outcome (0-3)	Total Score (0-9)	Comments
Brouns <i>et al.</i> , 2016	3	1	3	7	Selection bias due to specific setting, adequate follow-up but potential measurement bias.
Yang <i>et al.</i> , 2015	2	1	1	5	Significant selection bias, insufficient control for confounders, incomplete follow-up.
Chai <i>et al.</i> , 2021	3	1	2	6	Feasibility well assessed but potential selection bias and moderate follow-up.
Ward <i>et al.</i> , 2021	2	1	2	5	Some selection bias, adequate follow-up but with moderate outcome assessment.
Miller <i>et al.</i> , 2020	1	1	2	4	Effective treatment times, but potential selection and measurement biases.
Swanson <i>et al.</i> , 2021	3	1	2	5	Well-documented outcomes but poor follow-up and selection bias and potential for measurement bias.
Kadar <i>et al.</i> , 2019	1	1	2	4	Robust exposure and follow-up but potential selection bias, and lack of representativeness.
Tripod <i>et al.</i> , 2020	2	1	2	5	Representative population, but with lack of selection criteria and moderate outcome assessment.



Figure 1: Newcastle-Ottawa score summary, including sources of bias.

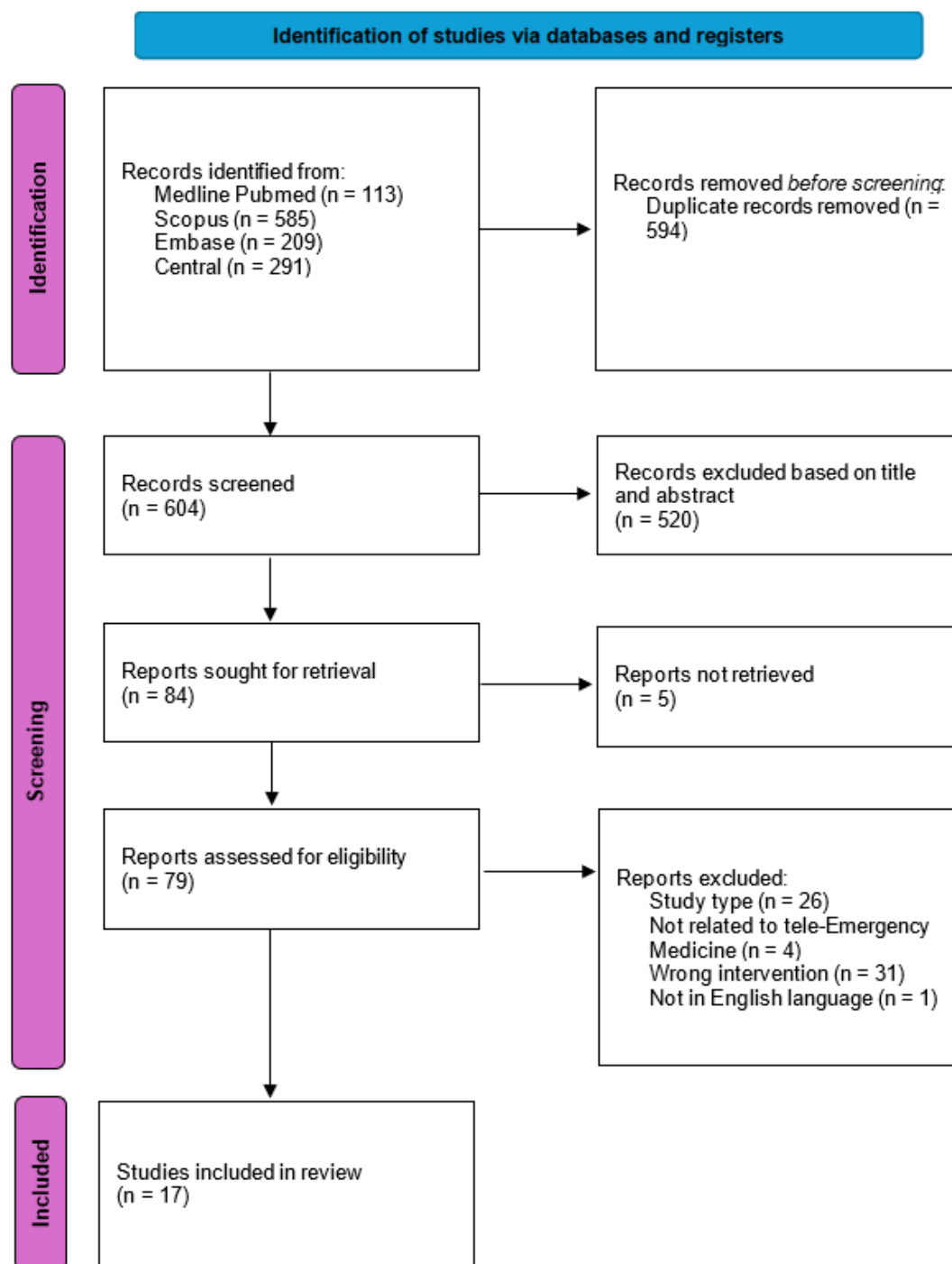


Figure 2: Identification, review and selection of articles included in the systematic review of telemedicine in Emergency Medicine

## **Diet and exercise interventions to promote cardiovascular health parameters in people with severe mental illness and first episode psychosis**

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### **Abstract**

#### *Aims*

Individuals with severe mental illnesses (SMI) such as schizophrenia are prone to multiple health problems, both physically and psychologically. Cardiovascular diseases (CVD) have a higher prevalence and risk in those with SMI, with CVD being the most common cause of death in SMI.

This review aims to explore the current literature on diet and/or exercise interventions employed for people with SMI and the impact these interventions have on cardiovascular health using parameters such as weight, BMI and inflammatory markers.

#### *Methods*

This review was carried out from September 2024 – February 2025, by searching the 'Pubmed', 'Scopus' and 'Cochrane Library' databases. Studies published in the last 15 years pertaining to diet and exercise interventions were included.

#### *Results*

Sixteen studies have met inclusion criteria. Dietary and nutritional advice and exercise were utilised either in tandem, or individually. Motivational coaching was employed in all interventions. Positive effects were generally seen in cardiovascular health parameters. 44% and 31% of studies showed optimistic changes in weight and BMI. Additionally, 25% and 19% of studies showed decreases in waist circumference and inflammatory markers respectively.

#### *Discussion*

Interventions that enrolled service users in programs that comprised of dietary and exercise support have positive impacts on cardiovascular health parameters, however intervention

length could be longer. Future programmes could have a longer running times to adequately analyse the health impacts.

## Use of Neuroendoscopy for Metastatic Brain Tumours: A Systematic Review

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### Abstract

#### *Introduction*

Neuroendoscopy enables biopsy and resection of metastatic brain lesions with minimal invasion. This review surveys evidence on neuroendoscopic techniques for such lesions.

#### *Methods*

We conducted a systematic literature search following the Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) method.

#### *Results*

Our search found 38 studies, with 32 involving 150 participants. Average age was 57 (range 37–80), with a female-to-male ratio of 51:54. Common primary sites included lung (42/113), breast (18/113), and melanoma (16/113). Supratentorial tumors accounted for 93 of 115 cases. Among 34 studies, procedures aimed for resection in 23, biopsy in 6 (3 with ventriculostomy), and both in 5. Resection extent was reported for 93 participants: gross in 60, near in 13, and subtotal in 20 cases. Thirteen studies with 34 participants noted 28 (82%) showed postoperative symptomatic improvement. Overall survival, reported for 50 patients across 4 studies, averaged 12.8 months.

#### *Discussion*

This is the first comprehensive systematic review of neuroendoscopy for brain metastases. Findings suggest promise, but limited evidence quality and quantity prevent firm conclusions.

## **AI-guided cardiovascular focused ultrasound by scanners without clinical or imaging experience: Ability to detect left ventricular structural and functional abnormalities**

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### **Abstract**

#### *Aims*

This study compared diagnostic performance of artificial intelligence (AI)-assisted acquisition of a focused cardiac ultrasound (FOCUS) by novice users to a formal echocardiogram to assess left ventricular (LV) size and ejection fraction (EF).

#### *Methods*

We prospectively enrolled 498 adults referred for a diagnostic echocardiogram. Novice scanners underwent a 4-hour introductory imaging workshop, then performed FOCUS using a point-of-care device (Philips Lumify) utilizing real-time AI-assisted image acquisition (Ultrasight). Images were assessed by AI-analytic software (Mayo Clinic), and two expert blinded echocardiologists. Low LVEF was defined as < 50% and LV dilatation by standard criteria.

#### *Results*

Median age was 67 (interquartile-range 56-75), 39% female, and body mass index range of 17-57 kg/m<sup>2</sup>. 101 (20.3%) exhibited LV dilatation, while 65 (13.0%) had low LVEF. The median scan time was 4 mins (IQR 3-5). Adequate views for AI assessment were achieved in 95.0% of cases and 97.4% for expert analysis. In a combined endpoint of low LVEF or dilated LV, the PPVs were 75% and 89-95%, and the NPVs were 86% and 89-94% for AI echocardiography and expert interpretations, respectively.

#### *Discussion*

AI-guided technology enabled users without imaging experience to successfully acquire images to assess LV size and EF accurately. While expert review improved the results, the AI-

derived findings demonstrated strong predictive values. This technology shows promise for facilitating low-cost community screening for asymptomatic cardiac dysfunction.

## **A systematic review of age-related macular degeneration in Asians**

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### **Abstract**

#### *Aims*

Age-related macular degeneration (AMD) is a leading cause of vision loss among the elderly globally. Despite the increasing prevalence of AMD in Asia, comprehensive studies on these populations remain limited. This systematic review aimed to consolidate current knowledge on AMD in Asians.

#### *Methods*

Three databases (PubMed, Embase, and Cochrane) were searched for studies published between 2014 and 2024, guided by the PRISMA guidelines. From 902 identified papers, 25 studies were selected.

#### *Results*

The prevalence rates of AMD across various Asian countries ranged from 4.90% to 7.00% for early AMD and 0.10% to 0.76% for late AMD. Risk factors consistently identified across studies included age, smoking, and genetic predispositions. Notably, a study highlighted the association between specific genetic polymorphisms and AMD in a Chinese population. Clinical manifestations in Asians differed from Western populations, with polypoidal choroidal vasculopathy (PCV) more frequently observed. Anti-VEGF (vascular endothelial growth factor) treatment remains the primary approach for neovascular AMD. This includes subtypes like PCV that are prevalent in Asians. Furthermore, photodynamic therapy (PDT) combined with anti-VEGF treatment showed significant efficacy in managing PCV, improving visual outcomes and polyp regression.

#### *Discussion*

This review emphasizes the need for tailored diagnostic and management strategies for distinct subpopulations.

## Effects of PRC2 loss-of-function on amino acid metabolism of paediatric AML cell lines

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### Abstract

#### *Aims*

This study investigates the effect of Polycomb Repressive Complex 2 (PRC2) loss-of-function on amino acid (AA) metabolism in paediatric Acute Myeloid Leukaemia (AML) cell lines. Given that PRC2 is altered in 15% of AML cases and contributes to poor prognosis, this study aims to identify specific AA dependencies that could highlight novel therapeutic targets in PRC2-mutated AML.

#### *Methods*

PRC2 haploinsufficient AML cell lines were subjected to AA depletion. Viability was assessed using trypan blue exclusion and alamar blue assays. Cell cycle progression was analysed using propidium iodide, while apoptosis was evaluated via western blotting for cleaved caspase markers.

#### *Results*

Compared to wild-type cells, PRC2 loss-of-function cells exhibited higher viability under cysteine depletion. Under glutamine depletion, cell cycle analysis showed that most PRC2 loss-of-function cells transitioned to and remained in the S-phase, while wild-type cells did not.

#### *Discussion*

PRC2 haploinsufficient cells demonstrate enhanced adaptability to specific AA depletions, particularly cysteine and glutamine. The observed S-phase progression and arrest suggest a potential mechanism for survival under metabolic stress, which could translate to a selective advantage in vivo. These findings highlight S-phase arrest as a potential therapeutic

vulnerability in EZH2-mutated AML, warranting further investigation into altered AA metabolic pathways.

## **Efficacy of Preoperative Lugol's Solution in Total Thyroidectomy for Graves' Disease: A Systematic Review and Meta-Analysis**

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### **Abstract**

#### *Aims*

Lugol's solution (LS) is administered preoperatively in patients with Graves' disease (GD) undergoing surgery to reduce vascularity and the risk of postoperative complications. This meta-analysis investigates the clinical impact of LS in patients with GD undergoing total thyroidectomy.

#### *Methods*

Ovid MEDLINE®, CINAHL, Ovid EMBASE, and the Cochrane Library were searched from inception by two independent reviewers using the preferred reporting items for systematic reviews and meta-analyses (PRISMA) guidelines. The review was registered prospectively on the PROSPERO database (CRD42024585797).

#### *Results*

From the 2243 studies screened, 11 met the eligibility criteria, with a total of 2257 patients with GD undergoing total thyroidectomy with LS (n = 760, 33.7%) or without (n = 1497, 66.3%). The use of LS did not show better outcomes compared to the control group regarding

intraoperative blood loss (MD = -24.6 mL, 95% CI: -56.2, 7.0;  $I^2 = 82\%$ ), endocrine-related injuries (OR = 0.85, 95% CI: 0.42, 1.74;  $I^2 = 76\%$ ), hypocalcaemia (OR = 1.11, 95% CI: 0.65, 1.91;  $I^2 = 61\%$ ), hematoma (OR = 1.44, 95% CI: 0.39, 5.29;  $I^2 = 58\%$ ), and recurrent laryngeal nerve injuries (OR = 0.70, 95% CI: 0.26, 1.93;  $I^2 = 53\%$ ).

### *Discussion*

This meta-analysis demonstrates that preoperative LS and control groups had similar clinical outcomes and safety in total thyroidectomy for GD, with benefits of LS to the general population remaining uncertain. Future randomized controlled trials with larger sample sizes and more robust selection criteria are necessary to ensure homogeneity between groups and to accurately measure the influence of preoperative LS in patients with GD undergoing total thyroidectomy.



## Trouble in Trials – Our Experience in Navigating Challenges with Clinical Research

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### Abstract

#### *Aims*

There is an increasing emphasis for medical students to get involved in research during their medical education. We aim to identify the challenges associated with clinical database research for first-time researchers as well as increase awareness of how the challenges may influence research outcomes and result interpretation.

#### *Methods*

We report the challenges faced when conducting a systematic review of 18 RCTs surrounding motor function measures and cerebral palsy in children obtained from 11 clinical trial databases.

#### *Results*

A key difficulty encountered was the inconsistency in search results, particularly with the Brazilian Clinical Trials Registry (REBEC). Inconsistencies in search filters between databases, discrepancies in the advanced search algorithms, technical website malfunctions lead to the search strategy being conducted multiple times with the most frequent number of trials and the statistical mode being used to obtain data.

#### *Discussion*

There should be an increased awareness of the challenges encountered in searching clinical databases in order for technological updates and algorithms developments to take place. In the future, a platform for medical students to share and learn experience should be developed in order to foster experiential learning and collaboration.

## **Oral health-related quality of life (OHRQoL) and its association with disease severity in oral lichen planus (OLP) patients: A cross-sectional study**

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### *Aims*

To examine OHRQoL and its association with disease severity in OLP patients, using the Oral Disease Severity Score (ODSS) as a measure of OLP disease severity

### *Methods*

64 OLP patients were recruited for this cross-sectional study at the Oral Medicine Clinic at Cork University Dental School and Hospital. The 15-item Chronic Oral Mucosal Disease Questionnaire (COMDQ-15) was used to assess OHRQoL. Participants were assessed clinically and disease severity was determined according to the Oral Disease Severity Score (ODSS). Univariate analysis was performed to assess the impact of OLP on OHRQoL and to measure association between OLP disease severity and OHRQoL. P values <0.05 were statistically significant.

### *Results*

All 64 participants were included in the final study sample. 23.4% were male (n=15) and 76.6% were female (n=49). All participants were white (n=64). Mean participant age was 62.98 ± 10.874. Women reported significantly worse OHRQoL than men in the Medications and Treatment domain of COMDQ-15 (p=0.047), in the Social and Emotional domain (p=0.015) and overall (p=0.034). A moderate positive correlation was observed between OLP disease severity and OHRQoL (r=0.531, p<0.001).

### *Discussion*

Clinicians may expect to observe poorer OHRQoL amongst female OLP patients. However, further research with equal gender representation is needed to corroborate this finding. OHRQoL in OLP patients is associated with their level of disease severity.

## Investigating the utility of BH3 mimetics in Oesophageal Adenocarcinoma (OAC)

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3. Department of Upper Gastrointestinal Surgery, St James Cancer Institute, Dublin, Ireland.

### Abstract

#### *Aims*

Oesophageal adenocarcinoma (OAC) is a highly prevalent and fatal cancer, with 5-year survival rates of 23-29%, largely due to chemoresistance. Defective intrinsic apoptotic mechanisms contribute to this resistance, with upregulation of anti-apoptotic proteins correlating with poorer survival. Cells resistant to apoptosis exhibit reduced priming, rendering them less responsive to chemotherapy. BH3 mimetics exploit this therapeutic vulnerability by enhancing apoptotic priming and increasing chemosensitivity in OAC.

#### *Methods*

An OAC cell line (OE19) was BH3 profiled to determine its apoptotic priming relative to other cell lines, using highly primed haematological cancer cell lines and unprimed non-cancer normal cell lines as comparators. Additionally, we investigated whether OE19 cells relied on specific anti-apoptotic proteins for survival, and if targeting these anti-apoptotic dependencies with BH3 mimetics could increase the apoptotic sensitivity of OE19 cells.

#### *Results*

OE19 cells exhibited low apoptotic priming, consistent with OAC's chemoresistant phenotype. Dependence on Bcl-xL, Bcl-2, and Bcl-w for survival was identified. Treatment with A1331852 (Bcl-xL inhibitor) and ABT-263 (Bcl-xL, Bcl-2, and Bcl-w inhibitor) significantly increased apoptotic sensitivity compared to DMSO controls.

#### *Discussion*

These results support the hypothesis of OE19 cells being unprimed for apoptosis and the ability of BH3 mimetics to exploit anti-apoptotic dependencies. Future research should examine the ability of ABT-263 and A1331852 in enhancing the efficacy of standard-of-care chemotherapy regimens in OAC.

## **Investigation of clinical risk factors related to the development of moderate and severe dysphagia following anterior cervical discectomy and fusion (ACDF) spine surgery**

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3. Student Summer Research Awards Program, UCD School of Medicine and Medical Science, Belfield, Dublin 4

### **Abstract**

#### *Aims*

Dysphagia is the most common complication following Anterior Cervical Discectomy and Fusion (ACDF) surgery, with both patient-specific and intraoperative factors contributing to its prevalence and severity. This study aims to identify potential risk factors related to the development of moderate and severe dysphagia in patients following ACDF surgery.

#### *Methods*

A retrospective chart review was conducted in Mater Misericordiae University Hospital of patients who had undergone ACDF surgery and had subsequent postoperative Fiberoptic Evaluation of Swallowing. Patients were stratified into cohorts based on dysphagia severity using the AusTOMS Score. Study variables collected included demographic factors, patient comorbidities, patient lifestyle factors, mechanisms of injury and surgical variables.

#### *Results*

A total of 53 patients were included, with a male predominance (36 males, 17 females). The mean age was 59.7 years for mild dysphagia, 65.0 years for moderate dysphagia, and 64.8 years for severe dysphagia. Active alcohol consumption ( $p = 0.04$ ), C2–3 level injury ( $p = 0.02$ ),

and postoperative complications ( $p < 0.01$ ) were identified as significant risk factors for severe dysphagia.

### *Discussion*

Severe, but not moderate, postoperative dysphagia following ACDF is influenced by patient and surgical risk factors. Standardizing dysphagia assessment and controlling for confounders could enhance understanding of its pathophysiology. Future research should explore targeted postoperative interventions, such as laryngeal rehabilitation, in high-risk patients to mitigate dysphagia severity.

## Identification of frailty and pre-frailty syndrome in People with HIV (PWH)

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### Abstract

#### *Aims*

This study aimed to determine the prevalence of frailty and pre-frailty phenotypes, physical activity and the degree to which patients had been investigated for other frailty markers in treated PLWH attending St. Vincent's University Hospital.

#### *Methods*

A cross-sectional study of virally suppressed PLWH enrolled from the All-Ireland Infectious Diseases (AIID) cohort was conducted. Recruited participants were assessed for frailty via the Fried Frailty Criteria and physical activity via the Rapid Assessment of Physical Activity (RAPA) Tool. Clinical and demographic data was also collected, notably FRAX scores.

#### *Results*

55 participants were recruited to this study with a median age of 38. Overall, 5.45%(3) met the criteria for frailty and 52.72%(29) for prefrailty. Of frail and pre-frail patients only 59.4%(19) had Frax screening scores. The most common characteristic of frailty seen was Low Physical Activity with 38.89%(21) of total patients matching the criterion. 80.39%(41) of all characteristics matched was accounted for by Poor Endurance and Energy and Low Physical Activity. 47.27%(26) of patients were classed as suboptimal on the RAPA assessment.

### *Discussion*

This study is the first frailty assessment carried out on the AIID cohort and results show high levels of Pre-frailty with low levels of activity and energy being particularly prevalent. This study also shows there's significant scope for further Frax screening for fracture risk in frail and prefrail patients.

## **Development of a Standardised Dataset for Referral of Complex Skin Malignancies for Adjuvant Radiation Treatment**

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### **Abstract**

#### *Aims*

Referrals for radiation oncology from plastic surgery and dermatology services often involve complex cases. Therefore, the inclusion of accurate visual documentation may facilitate precise treatment planning. This audit aimed to evaluate referral patterns, focusing on the inclusion of clinical photography.

#### *Methods*

A retrospective audit of complex skin cancer referrals, from the Mater Misericordiae University Hospital skin multidisciplinary team meeting to radiation oncology, over a 12 month period between October 2023 and October 2024 was conducted. Data on the information provided at referral including; clinical photography of the index lesion, anatomical site affected and pathology, were collected.

#### *Results*

A total of 31 patients were referred during the period audited. Of these, 23 patients had initial surgical assessment. Eight patients had photos sent, including one after request, all of whom

had prior surgery. The most common referral pathology was squamous cell carcinoma. The most common referral anatomical site was the forehead.

### *Discussion*

Although clinical photography may be beneficial, most referrals lacked its inclusion. These findings suggest the need for clearer guidelines regarding referrals made to radiation oncology. A referral proforma will be developed, in conjunction with dermatology and radiation oncology to ensure data standardisation for all referrals. This will reduce the risk of a wrong or inaccurately estimated treatment field. The referral process will then be re-audited.



## Identification of patients with a diagnosis of COPD and optimisation of the management of those on chronic treatment in a primary care setting

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### Abstract

#### *Aims*

This project aimed to optimise the care of chronic obstructive pulmonary disease (COPD) patients in a local GP setting by following the Health Service Executive (HSE) national guidelines for management of COPD. It focused on four key areas: vaccination status (influenza, pneumococcal and COVID-19), smoking cessation, inhaler technique and self-management plans.

#### *Methods*

The quality improvement process involved 160 patients, with 68 meeting the criteria for inclusion. Two assessment tools, the COPD Assessment Test (CAT) and the Medical Research Council Dyspnoea (MRC) Scale, were used to evaluate patients' current management.

#### *Results*

Key findings included a wide variation in CAT scores, indicating that many patients were managing their condition well, but some required intervention. Vaccination status was suboptimal, with many patients not having received recommended vaccines. The smoking status of patients revealed that 39 (57%) were ex-smokers, and 17 (25%) were current smokers, with smoking cessation advice provided. Occupational exposure to harmful substances was reported by 37 (54%) patients. Furthermore, improvements in pharmacological therapy were recommended for three patients.

#### *Discussion*

Future work includes a re-evaluation to assess the effectiveness of the implemented changes in improving patient care, as measured by subsequent CAT and MRC scores. This project highlighted areas for improvement, including vaccination uptake, smoking cessation, and pharmacological treatment, all aimed at optimising COPD management in line with national guidelines.

## Epitranscriptomic Aberrations of Breast Cancer Brain Metastasis

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### Abstract

#### *Aim*

The fat mass and obesity-associated gene (FTO) has been linked to poorer patient survival and increased cell proliferation in breast cancer brain metastasis (BCBM). While FTO is known to act as a demethylase of N6-methyladenosine, a transcriptomic modification, its exact mechanism remains poorly understood. This study aims to elucidate the role of FTO by evaluating its binding to potential transcript targets, NPM1 and MYC, in a BCBM (T347) cell line.

#### *Methods*

FTO-bound RNA was extracted by RNA immunoprecipitation (RIP), followed by reverse transcription and qPCR to assess MYC and NPM1 mRNA binding to FTO. A positive control with SNRNP70 and its known target RNU1 was included. qPCR data were normalised to IgG (100%) to account for non-specific binding.

#### *Results*

The positive control antibody SNRNP70, targeting RNU1, demonstrated enrichment (239.93%) compared to IgG (100%), confirming the experiment's validity. In contrast, MYC and NPM1 showed fold enrichment values of 83% and 90%, respectively, indicating no significant binding to FTO.

### *Discussion*

The study's results, compared with parallel experiments, suggest that FTO transiently binds to its transcript targets, NPM1 and MYC. Given their roles in cell cycle regulation, variability in binding levels may be due to RIP being performed at different cell cycle stages across experiments.

## **Factors impacting primary eye care delivery in Sub-Saharan Africa**

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### **Abstract**

#### *Aims*

In Sub-Saharan Africa (SSA), strengthening primary eye care (PEC) is crucial toward expanding healthcare access. However, research on PEC in many countries remains limited. Therefore, this review aimed to analyze current PEC delivery strategies in Sub-Saharan Africa to draw out any common barriers and facilitators across different countries in the region.

#### *Methods*

Ovid MEDLINE, Embase, Scopus, Cochrane Central Library, and ProQuest Public Health were searched in February 2024. Quotes from included studies were extracted and collated into custom data sheets. Relevant results were then extracted and summarized according to the WHO Building Blocks framework for healthcare systems. The frequency of reported barriers and facilitators was quantified to develop descriptive statistics, and study quality was assessed using CASP and MMAT checklists.

#### *Results*

The search generated 119 studies, 11 of which were included. Two additional studies were found in the reference lists of included studies. Low clinical knowledge (n=9), healthcare practitioner shortages (n=8), and equipment shortages (n=8) were the three most-commonly identified barriers to effective PEC. Improving clinical training (n=8) and sustained government support (n=8) were the two most commonly-reported facilitators.

#### *Discussion*

Primary eye care delivery is a complex, multifactorial issue. By identifying key factors that affect the delivery of primary eye care in sub-Saharan Africa, this review may inform policies

and interventions aimed at enhancing PEC accessibility, workforce capacity, and health system resilience.

## Effect of Steroidogenic Acute Regulatory (StAR) Protein on islet amyloid-induced mitochondrial dysfunction in male and female mice

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### Abstract

#### Aims

Human islet amyloid polypeptide (hIAPP) aggregates in >90% of individuals with type 2 diabetes. Steroidogenic Acute Regulatory (StAR) protein, which transports cholesterol into mitochondria, is upregulated in amyloid-laden islets of hIAPP transgenic mice, contributing to mitochondrial dysfunction [1]. When fed a high fat diet, male mice develop islet amyloid, while females only do so once oophorectomized [2]. Whether this sexual dimorphism extends to StAR expression is unknown.  $\beta$ -cell StAR deletion in hIAPP transgenic mice may improve islet mitochondrial function under amyloid-forming conditions *in vitro*, potentially in a sex-dependent manner.

#### Methods

Islets from hIAPP transgenic mice of four genotypes ( $\beta$ -cell-specific StAR Null (StAR<sup>fl/fl</sup>.Ins1<sup>cre/+</sup>.hIAPP), Wild Type (WT), Cre-control, and fl/fl control) were cultured in glucose media for seven days. Amyloid was confirmed by immunohistochemistry, StAR expression quantified by RT-PCR and mitochondrial respiration assessed by Seahorse XF Analyzer.

#### Results

In males, StAR deletion enhanced mitochondrial respiration compared to WT ( $p < 0.05$ ). However, these differences were reduced compared to controls. Females showed no

significant differences among groups (Figure 1). Comparing sexes, male StAR null islets exhibited higher basal ( $p<0.05$ ), ATP-linked ( $p<0.01$ ), and maximal ( $p<0.01$ ) respiration than females, while these sex-differences were absent in the WT and controls.

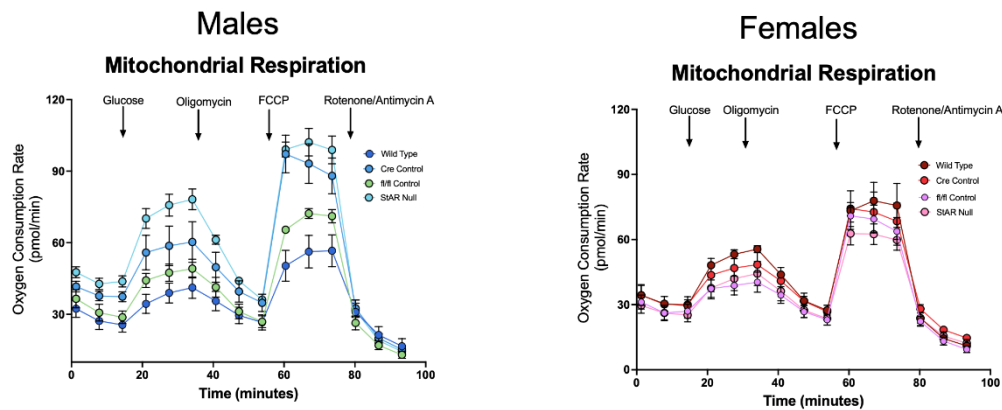


Figure 1: Measures of mitochondrial respiration in islets from male and female mice.

### Discussion

$\beta$ -cell StAR deletion improves mitochondrial function in male mouse islets. However, the role of fl/fl or Cre expression in this is unclear. Further, a sex-based dichotomy was observed, as female islets showed no response to StAR deletion - highlighting the importance of considering sex differences in mitochondrial studies.

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## Prospective and retrospective cohort study of diagnostic data in paediatric coeliac disease patients

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### *Aims*

Coeliac Disease (CD) may present at any age-group and is a common indication for endoscopic assessment. The aim of the study was to explore the demographic and diagnostic factors in paediatric patients with suspected CD.

### *Methods*

Patients were identified from the prospective SUCCEEDS study (Science Underlying Coeliac Evolution: Explanations, Discoveries, Solutions), from November 2022-January 2024. Retrospective data from 2018-2022 was collated and data analysed collectively. Serum levels of tissue transglutaminase IgA antibodies (tTg-IgA) were recorded in patients suspected of having CD, and diagnosis was confirmed through duodenal biopsy (Marsh Grade >2). The performance of tTg-IgA serology thresholds was assessed for diagnostic accuracy. Data were analysed using JAMOV statistical software.

### *Results*

A total of 883 patients were identified from the combined cohorts. Using serology results relative to the upper range of normal (ULN), values within 2.1-4x ULN had the highest sensitivity (90%), whereas values >10x ULN had the highest specificity (81%) and positive predictive value (90%). Youden's index was highest (0.29) at 4.1-6x ULN, with an area under receiving operating curve of 0.69. There were no significant predictors of CD ( $\chi^2$   $p > 0.05$ ).

### *Discussion*

Our real-world data suggest Irish coeliac serology testing performance is less than that quoted in published literature. Further research including analysis of follow-up data and recruitment of patients is necessary to interpret the data more accurately.

## Exploring the Interplay Between Asthma and Vitamin D: Insights from an Irish Population Study

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### Abstract

Vitamin D's anti-inflammatory properties may influence asthma control, though evidence is inconsistent. This study examined the link between vitamin D levels and asthma control in Irish adults.

### Methods

A retrospective review analysed the first 100 patients attending the CUH asthma clinic with Asthma Control Questionnaire (ACQ) scores and vitamin D levels, dating back from December 2022. Vitamin D was measured within  $\pm 30$  days of ACQ. From 195 data points (some patients had multiple sets), cross-sectional and longitudinal analyses assessed the vitamin D-asthma control relationship.

### Results

25% of patients had inadequate vitamin D, and 7% were deficient. Higher ACQ scores, indicating worse asthma control, were linked to inadequate vitamin D ( $P=0.04$ ). Longitudinal analysis showed no significant association between vitamin D changes and ACQ over time ( $P=0.33$ ). Lung function (FEV1) correlated with asthma control ( $P<0.001$ ), but not with vitamin D levels ( $P=0.66$ ). Vitamin D levels showed no seasonal variation, and supplementation did not significantly improve asthma control.

### Discussion

Inadequate vitamin D was associated with poorer asthma control cross-sectionally, but longitudinal vitamin D changes did not affect asthma outcomes. More research is needed to clarify the potential benefits of vitamin D supplementation for asthma management.

## Current state of Gene Therapy treatments for Beta Thalassemia

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### Abstract

#### *Aim*

To apprise clinicians about current gene therapy treatments for  $\beta$ -Thalassemia.

#### *Methods*

A literature review was performed based on ongoing clinical trials, systematic reviews, and long term follow up studies.

#### *Results*

$\beta$ -Thalassemia is a monogenetic hemoglobinopathy resulting in low adult haemoglobin (HbA) and accumulation of free  $\alpha$ -globin causing severe complications. Current treatment for the most severe form of  $\beta$ -Thalassemia includes regular blood transfusions and iron chelation therapy however, these are not curative and require lifelong treatment costing ~ \$5–5.7 million. Gene therapy offers a route to achieving curative results while significantly lowering long-term costs, and negating the risks of iron overload. *Zynteglo* and *Casgevy* are the first FDA approved gene therapy products for  $\beta$ -Thalassemia. *Zynteglo* transduces CD34+ hematopoietic stem cells with a functional  $\beta$ -globin gene allowing patients to become transfusion independent. *Casgevy* is a CRISPR-Cas9 therapy that reactivates fetal hemoglobin in patients, alleviating their symptoms.

#### *Discussion*

It remains of high interest to assess the effectiveness of implementing these treatments along with further evaluating whether they maintain beneficial effects long-term. Additionally, evaluating the accessibility of these treatments in developing countries where  $\beta$ -thalassemia



is highly prevalent is important. The continued efforts to provide gene therapies as a treatment option for patients is justified and encouraged by its potential to change the lives of those affected and greatly improve their quality of life.