

patient demographic data, ASA grade, surgical related complications, Recovery and histology results. We have extracted total time in theatre from theatre IT systems. We have looked into readmissions and return to theatre upto 30 days post surgery.

**Results:** The Audit is currently underway. Our Cohort include 250 cases over the last year of 2021. It is planned for completion by end of April 2022.

**Conclusion:** XXXX

## CLINICAL GENETICS

### OP.0049 | A 10-year review of antenatal skeletal anomalies in the pre-exome era

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**Objective:** Our objective was to evaluate diagnostic accuracy (including the added value of full post mortem) and pregnancy outcomes, in order to assist with improving antenatal counselling for families, and guide future service development in the genomic era.

**Design:** This was a retrospective cross-sectional study across our regional fetal medicine, neonatal and clinical genetics services.

**Method:** We searched our ultrasound reporting and image management database, for fetuses presenting with a skeletal anomaly between 2006 and 2017 ( $n = 119$ ), and excluded one for missing data ( $n = 118$ ). We recorded data from fetal-medicine ultrasound reporting, antenatal and postnatal clinic letters, post-mortem reports and genetic investigations. Analysis was descriptive, looking at skeletal and non-skeletal anomalies, yield of different diagnostic measures, and pregnancy outcomes.

**Results:** Most cases were identified at 19–25 weeks gestation. Short long bones was the most common presenting anomaly at 70% (83/118). The spectrum of phenotypes was broad; about half had one or more major non-skeletal congenital anomaly and about a quarter had severe prognosis due to thoracic/lung hypoplasia.

A clinical diagnosis was possible (although not always prenatally) in the majority of cases (97/118), of which about 40% (39/97) were genetically confirmed. Skeletal dysplasia was the most frequent diagnostic group ( $n = 38$ ), and thanatophoric dysplasia the most common amongst these ( $n = 10$ ). Other diagnostic groups were diverse and collectively more common. Karyotype, standard or molecular, was the most frequently performed genetic investigation. Targeted molecular testing had the highest diagnostic yield.

Termination was discussed in two-thirds of cases due to severity of prognosis; 21/76 declined, 11 of which resulted in a live birth, 5 survived to study date. Post-mortem was completed in 77 infants, which assisted in subtyping osteogenesis imperfecta and identified new diagnoses in 3 cases.

**Conclusions:** We identified a wide range of conditions and outcomes within this cohort. Concurrent non-skeletal major anomalies were frequent and often aided diagnosis and counselling. Most diagnoses were made on clinical presentation, antenatally and/or postnatally, with a subset confirmed genetically and a minority diagnosed at post-mortem. This suggests external examination alongside genetics and radiology may be an appropriate alternative to full post-mortem for some families. However, although we might expect genomic testing to increase diagnostic rate, this will also increase the chance of uncertain results that may need further details from post-mortems or family investigations for an accurate interpretation and feedback to families. Therefore, careful clinical phenotyping remains key.

### PP.0030 | Association of VDR gene variants with polycystic ovarian syndrome in Arab women

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**Objective:** To determine whether VDR gene variants are associated with polycystic ovarian syndrome (PCOS) in women of Arab ethnicity in Oman. Also to test the variant association with clinical and biochemical parameters.

**Design:** Prospective case control study.

**Materials and methods:** A sample size of 49 cases and 49 controls was estimated for a significance level of 5% at 60% power, with minor allele frequencies of 12% and 1% with a 1:1 ratio. We recruited 50 cases and 50 controls (Omani women) in reproductive age from the Gynecology clinic at Sultan Qaboos University Hospital in Oman. Cases fulfilled the Rotterdam criteria for PCOS. Controls were non-pregnant reproductive aged women who were not diagnosed with PCOS. Genetic disorders, congenital adrenal hyperplasia, androgen secreting tumors, Cushing syndrome, and hyperprolactinemia were excluded. Clinical and biochemical data was collected and analyzed. SPSS was used for statistical analysis. Gene Panel Exome Sequencing was conducted on DNA extracted from blood samples for VDR gene and variants were annotated with ANNOVAR software. Statistical analysis was performed using SPSS for Descriptive statistics, Chi square test and Correlation analysis. SNPs were tested for Hardy-Weinberg Equilibrium.

**Results:** After whole exome sequencing, only one nonsynonymous single nucleotide polymorphism (SNP) was identified which was VDR rs2228570. It was tested for association with different clinical biochemical and physical parameters among PCOS cases. The VDR rs2228570 SNP followed the Hardy-Weinberg Equilibrium (HWE) and there was no

association of VDR rs2228570 with PCOS. Cases with VDR rs2228570 (AG/GG) genotype compared to those that are (AA) genotype were found to have higher levels of testosterone (p-value 0.22), have more severe insulin resistance (p-value 0.95) and have higher levels of fasting blood sugar (p-value 0.66) but this did not reach statistical significance probably due to the small sample size.

**Conclusions:** VDR rs2228570 SNP was not found to be associated with PCOS in women of Arab ethnicity from Oman although there was a trend of higher levels of testosterone, fasting blood sugar and insulin levels in the AG/GG genotype cases compared to those that have AA genotype.

## EARLY PREGNANCY COMPLICATIONS AND GYNAECOLOGY

OP.0050 | abstract withdrawn

### OP.0051 | Reliability and usability assessment of Fine Birth device for TPTL diagnosis

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**Objective:** This article seeks to assess the reliability and usability of a novel medical device, the *Fine Birth*, aimed at accurately diagnosing Threatened Preterm Labor (TPTL) through the objective quantification of pregnant women's cervical consistency. Moreover, the present study evaluates the impact of a lateral micro camera on the device's reliability and usability outcomes.

**Design:** The proposed study is a clinical investigation with a medical device without CE marking, multicenter, prospective, non-randomized, and non-interventional in standard hospital care settings that aims to prove the usability of *Fine Birth* device and the reliability of the cervical consistency measurements.

**Methods:** 77 singleton pregnant women were recruited during their follow-up visit to the obstetric and gynecological services of 5 Spanish hospitals. Cervical consistency has been measured in each subject until obtaining two valid measurements by two different operators. The intra-observer and inter-observer reliability of the *Fine Birth* measurements were assessed using the Intraclass Correlation Coefficients with 95% of confidence interval, and the F-test p-value. In addition, the usability was also evaluated using variables based on clinicians' and recruited patients' use and feedback.

**Results:** The intra-observer reliability results obtained showed a clear concordance between single operator measurements (ICC>0.8 and F-test p-value <0.05). Since the results obtained for the inter-rater reliability did not reach the desired acceptable values, a lateral microcamera was added to the *Fine Birth* intravaginal probe, and the operators involved in the clinical investigation received the corresponding training with the modified device. The statistical analysis of 16 additional recruited subjects demonstrated a clear improvement of the inter-rater reliability results after the modification.

**Conclusions:** The robust reliability and usability results obtained after the insertion of a lateral micro camera and the training perform with the modified device, make *Fine Birth* a promising novel device to objectively quantify the patient's cervical consistency, diagnose TPTL and thus, predict the risk of spontaneous preterm birth.

### OP.0068 | Predicting tubal ectopic pregnancy and miscarriage in serum using Fourier transform-infrared spectroscopy

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**Objective:** Ectopic pregnancy and miscarriage are associated with maternal morbidity and mortality. Current management of pregnancy of unknown location and viability may involve serial blood human chorionic gonadotrophin-beta measurements and several ultrasound scans. Multiple interventions and length of time to diagnosis are costly and have a profound psychological impact on women. Vibrational spectroscopic methods such as Fourier transform-infrared (FT-IR) spectroscopy have been increasingly shown to discriminate disease states such as cancer from controls utilising the 'metabolomic signature' of molecules within a biological sample. It is hypothesised that this signature will differ between live normally-sited pregnancies, and tubal ectopic pregnancy and miscarriage.

**Design:** The Ectopic Pregnancy Diagnosis sTudy (ExPeDITE) was a prospective cohort study. In total, 380 pregnant women ≥18 years presenting with pain and/or bleeding at <10 weeks of gestation were recruited between November 2018 and November 2021. A blood sample and demographic data were collected at the time of consent, and a full history was taken. Early pregnancy outcomes were collected and classified according to ESHRE guidance.

**Method:** Harvested serum samples ( $n = 340$ ) were diluted 1:4 to prevent oversaturation of spectra and quality controls were created from the pooled diluted serum. Samples were transferred to silicon plates and dried at 37.5°C. Samples were subjected to FT-IR spectroscopy and spectral data were submitted to baseline correction, smoothing, and vector normalisation prior to multivariate statistical analysis