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ultrasound with evidence of ventriculomegaly and left diaphragmatic hernia. The examination performed at our institution confirmed these findings. The impression was that the stomach was inside the diaphragmatic hernia. In addition, there seemed to be corpus callosum agenesis. Fetal MRI was consistent with the ultrasound findings and confirmed the presence of corpus callosum agenesis. In addition to the stomach, the intestine was also intrathoracic. Fetal echocardiography showed only one heart deviated to the right. Fetal karyotype evidenced a structural abnormality in what appeared to be one of the 15 chromosomes. The child was born at 37 weeks of pregnancy, weighing 2,675 grams. She did not show any external abnormality or dysmorphism. She evolved with shock, hypoxemia and severe pulmonary hypertension and died within one day of life. The karyotypic evaluation of the mother showed that she had a translocation between chromosomes 8 and 15 [46,XX,t (8;15) (q11.21; q13)]. This result allowed us to define the alteration presented by the child: partial trisomy of chromosome 8 and partial monosomy of chromosome 15 secondary to a translocation of maternal origin [46,XY,+der (8) t (8;15) (q11.21; q13), -15 mat]. In our review, we did not find cases of diaphragmatic hernia associated with this chromosomal alteration. The identification of the balanced chromosomal abnormality in the mother was essential for the correct genetic counseling of the family.

VP16.21
**An unusual course of ductus venosus in a Trisomy 21 fetus**

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A 34-year-old patient was admitted for the first trimester scan. It was 13th week of her second pregnancy. The examination showed oedema of the subcutaneous tissue (the nuchal translucency was 6,6mm) and impaired ossification of the nasal bone. The right heart was enlarged, with tricuspid regurgitation, with apparently normal big vessels appearance. During examination of the ductus venosus an anastomosis between the descending aorta and the ductus was observed. The patient was offered chorionic villus sampling. The biopsy showed Trisomy 21. On the follow-up visit in the 16th week, the fetus was re-examined. We were surprised to see this time a normal course and normal flow of the ductus venosus. Meticulous examination of the heart showed no disproportion between left and right heart. Moreover, no tricuspid regurgitation was seen. In the fetal liver, an area of calcifications was noted. We hypothesised that calcifications formed when the anastomosis between the aorta and the ductus venosus closed. We also hypothesised that enlargement of the right heart and tricuspid regurgitation was due to the presence of the anastomosis. To our knowledge a malformation of this kind was not yet described.

Supporting information can be found in the online version of this abstract

## VP17: ADVANCED TECHNOLOGY FOR DECISION SUPPORT

VP17.01
**Exploring a new paradigm for the fetal anomaly ultrasound scan: artificial intelligence in real-time**

J. Matthew[4,1], E. Skelton[4,1], T. Day[4,1], V. Zimmer[4], A. Gomez[4], G. Wheeler[4], N. Toussaint[4], T. Liu[2], S. Budd[2], K. Lloyd[4], R. Wright[4], S. Deng[4], N. Ghavami[4], M. Sinclair[4], Q. Meng[2], B. Kainz[2,4], J. Schnabel[4,3], D. Rueckert[2,3], R. Razavi[4], J. Simpson[1,4], J. Hajnal[4]

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**Objectives:** Ultrasound (US) is characterised by high levels of operator subjectivity and variability. Advances in artificial intelligence (AI) have demonstrated the potential to reduce both. This study pilots the end-to-end automation of multiple elements of the mid-trimester obstetric screening US scan using AI-enabled tools.

**Methods:** A single centre, prospective method comparison study was conducted. Participants had both standard manual and AI-assisted US scans, each performed alternately and independently by 2 blinded sonographers. The AI tools automated the acquisition of standard plane images, measurements, and the production of a written clinical report with saved images available for review. A feedback survey captured the sonographers' perceptions of scanning.

**Results:** 23 subjects were studied. The average time saving per scan was 7.62min (34.7%) when using the AI-assisted method ($p < 0.0001$) with no difference in reporting time. There were no clinically significant differences in biometric measurements between methods. The AI tools saved a satisfactory view in 93% of the cases when considering the four core views only, and 73% for the full 13 views, compared to 98% for both using the manual scan. Survey responses suggest the AI tools helped sonographers to concentrate on image interpretation by removing disruptive recording and measurement tasks.

**Conclusions:** Using AI to automate tasks during the ultrasound examination changes workflow. Separating the process of freehand scanning from image capture and measurement resulted in a faster scan. Reducing the need for sonographers to focus on repetitive tasks may allow more attention to be directed towards identification of atypical fetal anatomy. Further work is required to improve the performance of the image plane detection algorithm for use in real-time. In the future, high-quality AI tools could allow the sonographer to increase their focus on anatomical assessment for congenital anomaly detection and provide higher-quality parent-centred care.

VP17.02
**Description and clinical validation of a real-time AI diagnostic companion for fetal ultrasound examination**

J. Stirnemann[1,5], R. Besson[2], E. Spaggiari[1], N. Bourgon[1], S. Rojo[2], F. Loge[2], H. Peyro-Saint-Paul[2], S. Allassonniere[3], E. Le Pennec[4], Y. Ville[1,5]

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**Objectives:** To describe a real-time decision support system (DSS), named SONIO, to assist ultrasound-based prenatal diagnosis and

to assess its performance using a clinical database of precisely phenotyped postmortem examinations.

**Methods:** This DSS is knowledge-based and comprises a dedicated thesaurus of 294 syndromes and diseases. It operates by suggesting, at each step of the ultrasound examination, the best next symptom to check for in order to optimise the diagnostic pathway to the smallest number of possible diagnoses. This assistant was tested on a single-centre database of 209 cases of postmortem phenotypes with a definite diagnosis. The primary outcome was a target concordance rate > 90% between the postmortem diagnosis and the top-7 diagnoses given by SONIO when providing the full phenotype as input. Secondary outcomes included concordance for the top-5 and top-3 diagnoses; We also assessed a "1-by-1" model, providing only the anomalies sequentially prompted by the system, mimicking the use of the software in a real-life clinical setting.

**Results:** The validation database covered 96 of the 294 (32.65%) syndromes and 79% of their overall prevalence in the SONIO thesaurus. SONIO failed to make the diagnosis on 7/209 cases. On average, each case displayed 6 anomalies, 3 of which were considered atypical for the condition. Using the 'full-phenotype' model, the success rate of the top-7 output of Sonio was 96.7% (202/209). This was 91.9% and 87.1% for the top-5 and top-3 outputs respectively. Using the "1-by-1" model, the correct diagnosis was within the top-7, top-5 and top-3 of SONIO's output in 79%, 73% and 68%.

**Conclusions:** Sonio is a robust DSS with a success-rate > 95% for top-7 ranking diagnoses when the full phenotype is provided, using a large database of noisy real data. The success rate of 79% using the '1-by-1' model was understandably lower, given that SONIO's sequential queries may not systematically cover the full phenotype.

VP17.03

## Prediction of low fetal fraction in maternal blood cell-free DNA using a machine learning model

G. Rizzo[1,2], I. Mappa[1], F. Restaldi[3], P. Maqina[1], A. Alessio[1], A. Novelli[3]

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**Objectives:** Although non-invasive prenatal testing (NIPT) based on cell-free fetal DNA fragments in maternal blood samples is a well-established method for aneuploidy screening, in approximately 4% of women NIPT fails to return a result due to an insufficient fetal fraction (FF) cell-free DNA in maternal plasma. Since identification of women at risk of NIPT failure is crucial in counseling, we evaluated the feasibility of using a machine learning (ML) model based on maternal and fetal characteristics to predict low FF.

**Methods:** This was a prospective study on women undergoing NIPT at 11+0–13+6 weeks of gestation at our institution from January 2018 to May 2021. Criteria of exclusion were multiple pregnancies, fetal structural and chromosomal anomalies. Different maternal and feto-placental features were considered. A multivariate logistic regression model was first constructed for the prediction of a FF < 4 % using the variables significantly different in univariate comparison. A ML gradient-boosting model was then built to assess the personalised likelihood for low FF. Validation of the robustness of the ML model was done by cross validation a process that involves splitting the dataset into training and test sets.

**Results:** Of the 2643 available patients 103 (3.9%) showed a FF < 4%. 1762 were allocated to the training set and 881 to the testing set. The most significant measures used by the model to predict low FF in a descending ranking order were body mass index, gestational age, placental size, parity, uterine Dopplers, fetal heart rate and medications. The final ML model' showed a receiver operating characteristic curve with an area under the curve (AUC) of 0.78 which was significantly higher (De Long $p < 0.01$) than those developed by multivariate logistic regression (AUC 0.71).

**Conclusions:** Application of a ML algorithm allows to assign a personalised risk score for having low FF at NIPT superior to the logistic approach. This may help in women counselling on the selection of the adequate procedure including diagnostic testing or alternative screening tests.

VP17.04

## Three-dimensional printing from prenatal ultrasonography for visually impaired expectant parents: an international protocol

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**Objectives:** The experience of pregnancy sets visually impaired expectant parents apart from nondisabled people for whom viewing of the first ultrasound has become a social and emotional milestone. Vision loss leads them to difficulties, such as pregnancy embodiment, mental representation, dependency upon oral description by health care providers, and early fetal attachment. In parallel to these difficulties, three-dimensional (3D) printing technology from surface-rendered prenatal sonographic views has recently allowed to better apprehend fetal abnormalities in nondisabled patient, improving their comprehension. Therefore, we proposed to improve the societal inclusion of visually impaired expectant parents, by using 3D printing technology.

**Methods:** 3D-printing of tactile models from ultrasonographic examinations between 12 and 28 gestational weeks were mainly used. To obtain better 3D printed results, the presence of amniotic fluid is indeed necessary around the fetal face. The analysis protocol was based on multiplanar reconstructions in the three traditional orthogonal planes and tomographic reconstructions. Fused deposition modelling technology was used to obtain the haptic models and multiple materials have been tested. Satisfaction was assessed by a satisfaction questionnaire performed orally at the end of reading the printed models.

**Results:** Prenatal tactile 3D ultrasonography therefore offers visually impaired persons a sensory vector that is useful for bridging the disability-related gap, allowing for societal inclusion. This tactile experience can enable an emotionally richer aspect to the medical consultation, offering impaired expectant parents an accommodation that allows the first ultrasound appointment to be refocused on the parents' encounter with their unborn child. They can thereby perform their own mental representation process by extrapolating sensory information obtained from 3D tactile support.

**Conclusions:** 3D printed models improve the sonographic experience of visually impaired expectant parents.

VP17.05

## Improved patient education for prenatal aneuploidy testing using a digital tool: a multicentre randomised controlled trial

R. Jones[4], K. Chen[4], E. Kostenko[4], A. De Maria[3], B. Su[4], A. Villa[2], M. Swarup[2], J. Weida[1], M.G. Tuuli[1]

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