cardiac abnormalities and genetic anomalies. The most frequent genetic abnormalities associated with ARSA are chromosomal aberrations, especially Trisomy 21 and Trisomy 18. We report a case of a mosaic Turner Syndrome detected by karyotype analysis performed due to ARSA as an isolated sonographic finding. A 28-year-old woman, primigravida, started pregnancy surveillance only at 22 weeks. The first ultrasound, at 23 weeks, revealed an ARSA, without other changes. After genetic counselling, the parents opted for karyotype analysis and FISH for 22q11 deletion that revealed a mosaic 45XO/46XX, with about 19% of the cells in monosomy. At 36th week of gestation, the estimated fetal weight was in the 4th percentile with abnormal Doppler blood flow in middle cerebral artery and umbilical artery. An induction of labour was decided a week later and an 2252gr female infant with Apgar score of 9/10 was delivered by vaginal delivery. Postnatal examination and echocardiography confirmed that ARSA was the only finding in the fetus.

Turner’s syndrome is a relatively common chromosomal disorder occurring in approximately 1 in 2000 live births. The syndrome is caused by complete or partial absence of the second X chromosome, with or without cell line mosaicism. The most serious clinical aspect of the syndrome is due to congenital cardiovascular anomalies, in particular elongation of the transverse arch and aort coarctation. Mosaic monosomy X has been associated with a significant lower incidence of major cardiovascular anomalies. The performance of a fetal karyotype test when ARSA is an isolated finding is still a debatable issue, however it should be discussed with the parents.

**VP32.04**

Prenatal diagnosis of a case of Jeune syndrome in combination with Down's syndrome

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Jeune syndrome is an extremely rare condition representing asphyxiating thoracic dystrophy (ATD) and it is potentially lethal congenital dwarfism with estimated incidence of 1 per 100,000–130,000 livebirths. We present a case of Down’s syndrome that has signs of skeletal dysplasia and was confirmed by prenatal genetic analysis at 19 weeks of gestation age. A 27-year-old woman, no consanguinity, has previous child that died after birth several day due to severe respiratory distress since birth along with multiple characteristic skeletal anomalies including small bell shaped thorax and short limbs. Follow-up of the mother in her second pregnancy with prenatal sonographic examination at 12 weeks with thick nuchal translucency 3mm and abberant right subclavian artery. Aminocentesis revealed Trisomy 21 at 16 weeks. Ultrasound before termination of pregnancy showed short long bones below 10 percentile, small bell-shaped chest (TC/AC = 0.72), dip at thoracic-abdominal junction, short horizontal ribs, echogenic foci in left ventricle, thick nuchal fold and prenasal skin without polydactyly and kidney abnormalities.

Exome sequencing revealed fetus has 3 mutation in DYNC2H1 gene: NM_001080463.2(DYNC2H1):c.6560A>G (p.Gly2187Gly), NM_001080463(DYNC2H1): c.10571G>A (p.Arg3524His), (Suppl. 1): 57–378.

**VP32.05**

Prenatal and postnatal findings of a fetal overgrowth syndrome with complex vascular anomalies: CLOVES Syndrome

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Overgrowth syndromes with complex vascular anomalies are rare and caused by mosaic mutations in the PIK3CA gene. Clinical features vary widely and usually include fat tissue overgrowth, vascular, skeletal and renal anomalies. We report the ultrasound features and perinatal management of a case of a congenital malformation, highly suggestive of CLOVES (Congenital Lipomatous Overgrowth, Vascular malformations, Epidermal nevi and Skeletal/Spinal anomalies) Syndrome, a rare non-hereditary syndrome. A 27-year-old, gravida 3 para 2, Polynesian woman was referred to the Feto-Maternal Unit (FMU) of Liverpool hospital at 21+3 weeks of gestation for further evaluation of an anomaly ultrasound showing cystic lesions around the fetal body. The follow-up ultrasound showed ascites and multiple subcutaneous cystic swellings of the torso and limbs. After multidisciplinary counselling, the couple requested termination of the pregnancy. The postnatal examination of the fetus revealed cystic anomalies, macrodactyly and sandal gap of the toes. Autopsy was declined. Fetal x-ray demonstrated swelling of the soft tissues and clindactyly of the great toes. The fluid aspirated from the cystic lesions was compatible with lymphatic origin of the lesions and was sent for cytogenetic evaluation. No abnormalities were found on microarray testing and the specific mutation analysis of the PIK3CA gene is still pending. Currently the patient is 19 weeks' pregnant with no ultrasound abnormalities detected. This case highlights ultrasound features that can help with the diagnosis of CLOVES syndrome in the prenatal period.

**VP32.06**

Early detection and differential diagnosis of syndromic craniosynostosis: a case series

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Syndromic craniosynostosis is a rare genetic condition caused by premature fusion of one or multiple cranial sutures combined with malformation of other organs. An early detection is important for interdisciplinary counselling of the parents and planning of perinatal management. We aim to investigate sonographic signs of different syndromic craniosynostoses as well as associated malformations to make a precise and early diagnosis. Using the data system Viewpoint, we identified in the period of 2000–2019 12 cases with a prenatal suspected diagnosis of syndromic craniosynostosis at the Department of Obstetrics, Charité Universitätsmedizin Berlin. We analysed the ultrasound findings, MRI scans, genetic results as well as the mode of delivery and postnatal procedures.

Eight children were diagnosed with Apert syndrome, two with Saethre-Chotzen syndrome and one with Crouzon syndrome and Greig cephalopolysyndactyly syndrome. The majority of those were detected in the second trimester screening. We identified characteristic changes of the head shape as well as further malformations typical for those syndromes. Syndactyly was the most common malformation and detected in nearly all cases. After the diagnosis, six parents decided to terminate the pregnancy. Of the other six children, five were delivered by Caesarean section and one by vaginal birth.
VP32.07  
Prenatal diagnosis of Caroli’s syndrome in association with autosomal recessive polycystic kidney disease  

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Carol’s disease/syndrome is a rare congenital anomaly characterised by non-obstructive saccular or fusiform multifocal segmental dilatation of the intrahepatic bile ducts. It is usually seen with autosomal recessive polycystic kidney disease (ARPKD), a rare ciliopathy with an incidence of 1:20000 livebirths.  

Prenatal diagnosis of the ARPKD associated Caroli’s disease/syndrome is rare. Herein, we present a case of a 29-week fetus with ARPKD associated with Caroli’s syndrome which has prenatally confirmed by ultrasound, MRI, and anamnestic, pathological and genetical findings after induction.

VP32.08  
Clinical case of pregnancy and follow-up of Bartter syndrome (type 2) with a novel mutation  

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Bartter syndrome is a rare autosomal recessive inherited salt-wasting tubulopathy.  

We represent the case report of a Bartter syndrome (type II) with a novel mutation in KCNJ1 gene: c.554C>T (p. Pro185Leu) from antenatal presentation to 6 months postpartum. 29-year-old pregnant woman with BMI 46 was referred to Riga Maternity Hospital due to severe polyhydramnios at 26 weeks of gestation. A detailed ultrasound examination revealed anatomically normal fetus. The fetal biometric data of the patient was consistent with 27/28 gestational weeks with polyhydramnios (AFI 36.4) and shortened cervix (7 mm). A normal oral glucose tolerance rule out gestational diabetes mellitus. MRI excluded severe malformations. Diagnostic amniocentesis and amnioreduction reduced maternal dyspnoea and excluded chromosomal abnormalities and TORCH infections. The amniotic fluid analysis results showed the low protein level (1.96 g/l), elevated sodium (144 mmol/l), and chlorides in the upper reference limit (108 mmol/l). These enabled suggest Bartter syndrome. At 32 + 2 weeks labour was induced due to maternal respiratory distress and wide choioamnionitic membrane separation. Labour was complicated by severe placental abruption and newborn – boy was referred to NICU. Neonatal period was complicated by electrolyte abnormalities: hyponatremia, hypochloremic metabolic alkalosis, transient hypercalcemia that gradually developed into hypokalemia, hypercalcemia and elevated renin and aldosterone levels characteristic to type II Bartter syndrome. At 6 months he is gaining weight within normal ranges and his psychomotor development is ahead of his corrected age, without any need for daily medications.  

The present case report describes the clinical course of a Bartter syndrome with novel mutation and its favorable prognosis for the neonate. Early recognition and interventions are essential to prolong the pregnancy and reduce complications of prematurity.

VP32.09  
Bilateral diaphragmatic hernia in a case of Cornelia de Lange syndrome  

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A 38-year-old was sent to our unit for the first trimester scan at 12 weeks pregnancy. The first trimester calculated risk for trisomies was high; b-HCG-0.6 and PAPP-A-0.3. Subsequent amniocentesis revealed normal catytype 46 XY. Array-comparative genomic hybridisation study didn’t reveal any abnormality as well. Our scanning at 12 and 20 weeks revealed severe micrognathia (at 12 weeks) with mandibular spur (20 weeks), dysmorphic facial features such as hypertelorism, long convex philtrum, antverted nares, depressed nasal bridge, thin lips with downturned lip corners at 20 weeks. Scanning at 20 weeks also revealed bilateral diaphragmatic hernia, stomach and bowel loops located in the left hemithorax, liver located in the right and left hemithorax, anterior displacement of the heart with minimal lateral shift, diaphragm was visible only at its anterior part, small hands with brachydactyly, cleft palate was suspected. After TOP the WES test revealed heterozygous mutation in exon 10 in the NIPBL gene resulting in a frameshift mutation, which is associated with Cornelia de Lange Syndrome. Postmortem appearance of the abortus revealed also bushy eyebrows with synophrys, small hands and cleft palate. Diaphragm was presented only at its anterior part. Bilateral diaphragmatic hernia was confirmed.

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

VP32.10  
Identification of a de novo mutation of ENG gene in a fetus with pulmonary arteriovenous fistula in a Chinese family  

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Pulmonary arteriovenous fistula or malformation (PAVM) is the direct communication of pulmonary artery system or venous system bypassing the capillary. PAVM is a progressive disease associated with hereditary hemorrhagic telangiectasia (HHT) and it’s rarely seen in fetus. Prenatal ultrasound examination has been performed on the pregnant patient. Magnetic resonance imaging (MRI) has been performed on the aborted fetus. Clinical whole-exome sequencing (WES)-trio test were conducted in the family members including fetus, father and mother.  

Here we report on a 31-gestational week pregnant woman prenatally diagnosed with PAVM through ultrasound examination. The pregnant patient and her husband chose to abort the fetus. PAVM has been confirmed by MRI on the aborted fetus with consent of parents. WES-trio result show a novel de novo mutation ENG, c.245_246delTC (p.Leu82Profs*66) was identified in the fetus.  

In conclusion, a novel de novo mutation of ENG was identified in a PAVM fetus. Contrary to conventional perceptions, the causative gene of HHT can lead to fetal PAVM.