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# World Congress of **Perinatal Medicine**

# ISTANBUL - 2019 11-14 September

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Perinatal Medicine Foundation
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## ABSTRACT BOOK

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# ABSTRACT BOOK

# SPEAKERS

## Abstracts

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## E1407 - ADVANCES IN PRENATAL DIAGNOSIS AND DECISION TO TERMINATE PREGNANCY AT UNIVERSITY CLINIC OF GYNECOLOGY AND OBSTETRICS

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### Objective

The purpose of prenatal diagnosis is to obtain the appropriate information regarding prognosis, risk of recurrence, and potential therapy, and thus achieve optimum outcome for the fetus and the mother. Making a decision to terminate pregnancy is always on the parents' indication that the fetus with an increased nuchal translucency may have a normal karyotype at the start of diagnosis, a range of malformations that may develop later in pregnancy and after screening for organic malformations of 20-24 gestational week, as well as a long list of explanations for possible consequences in relation to the fetus if pregnancy is not terminated.

### Methods

In the article, interruptions of pregnancies were considered during one year, 2017, at the Clinic for Gynecology and Obstetrics, the same systematized after gestation weeks. Before any termination of pregnancy amniocentesis was performed in the clinic and all other diagnostic examinations according to the week that are within the scope of the Clinic. The purpose of the review is to begin the termination of pregnancy, determined using diagnostic facilities before the interruption. Fetal anomalies divided into four major groups: cardiac defects, nervous system defects, chromosomopathies, others (covering abdominal wall defects, urinary system, hydrops, dysplasia, IVGR, multiple anomalies).

### Results

According to the data on termination of pregnancy at the University Clinic of Gynecology and Obstetrics, in 2017, the total number of interruptions of pregnancy is greatest in the period from 20.1 to 24 gestational week, or 44% of all interruptions of pregnancy. Aborted pregnancies above 28 weeks of gestation are 11%, most of them due to defects in the nervous system (34%), and equally represented cardiac defects and chromosomopathies. Neural tube defects (anencephaly, spina bifida, cephalocele, holoprosencephalus, acrony ...) are the second most common fetal abnormalities after cardiac anomalies (Williams, Cragan, 2009, Dolk, 2010), or interruption of pregnancy due to the same in our examined group 40% of all interruptions over one year. Discontinuation of pregnancy due to a cardiac defect in the examined group is 10% of all interruptions in a year, due to cardiopathia complex.

### Conclusion

The determination of Nuchal translucency and its elevated values are only the beginning which points to additional diagnostics and confirmation of fetal abnormalities. Further biochemical screening in the second trimester, ultrasound echocardiography and screening for structural defects of other organs and organ systems complement the diagnosis of fetal abnormalities in subsequent weeks of pregnancy, unless an abortion occurred in early pregnancy or fetus in utero death. Amniocentesis affects parents' decision-making, their desire for prolonged pregnancy, or a decision to prolong pregnancy with otherwise unhealthy organic anomalies and heart failure, which at some point in the pregnancy would end up in fetal death or soon after birth. Reduction of the percentage of interruption of pregnancy due to anomalies that are the first in the presence of the fetus is due to the progress in the operative resolution of the fetus.

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## E1418 - NOONAN SYNDROME ANTENATAL DIAGNOSIS AND CLINICAL IMPLICATIONS

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**Objective**
Noonan syndrome is a rare genetic disorder with its prevalence estimated between 1 in 1,000 to 2,500 live births, affecting equally male and female infants. It is an autosomal dominant disorder mainly, yet both autosomal recessive inheritance and de novo cases have been reported. The syndrome presents a clinical and genetic heterogeneity, characterized by short stature, mild facial malformations, congenital heart abnormalities, and a variety of cognitive deficits. Mutations in seven genes have so far been associated with Noonan syndrome, especially those related to the PTPN11 gene. These mutations are thought to cause dysregulation in the RAS-MAPK signaling pathway, which also appears to be involved in the pathogenesis of other syndromes such as Leopard syndrome (LS), Noonan-like(with loose anagen hair, NS/LAH) syndrome, Costello's syndrome (CS), Cardio-Facio-Cutaneous syndrome (CFCS), Neurofibromatosis Type I (NF1) and Legius syndrome (LS). We present 3 cases of Noonan's syndrome, that were diagnosed antenatally as a result of detection of suspicious ultrasonographic findings in the fisrt trimester of pregnancy, and discuss the role of detailed screening by a specialist in fetal medicine, as well as the multiple challenges of a comprehensive parental counseling, once the diagnosis has been confirmed by molecular karyotype.

**Methods**
In the first trimester scan of 12-13 weeks, performed in a fetal-maternal referral unit, sonographic findings such as cystic hygroma in one case and increased nuchal translucency in two cases, lead to Chorionic Villous Sampling (CVS).

**Results**
In all 3 cases, molecular karyotype revealed the characteristic PTPN11 gene mutation suggestive of Noonan syndrome. An extensive parental counseling by a specialist in fetal-maternal medicine, as well as by a pediatrician with expertise in congenital malformations and genetic disorders followed, regarding the prognosis, the clinical heterogeneity and the broad spectrum of manifestation of signs and symptoms, the treatment options available after birth, the implications on future pregnancies and the need for genetic testing of both parents. All couples decided to terminate pregnancy, referring as their major concern the possibility of a severe cognitive disorder.

**Conclusion**
Diagnosis of Noonan Syndrome is mainly based on genetic testing, however, antenatally, due to absence of specific ultrasound findings, it should be suspected in any case of first trimester increased nuchal translucency or cystic hygroma, or increased or persistent nuchal fold during the second trimester of pregnancy. The role of ultrasonographic screening is indispensable, especially in the absence of a family history, to guide further testing to confirm diagnosis, counseling, clinical management and future implications.