



Conference Report

# 12th ISNS European Regional Meeting Oral and Poster Abstracts

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**Abstract:** Due to the impact worldwide of COVID-19, the 12th European ISNS meeting planned to be live in Luxembourg in November 2020 became Luxembourg Going Virtual in November 2021. The conference theme derived from the geographic location of Luxembourg was retained: *Newborn screening—working together in the heart of Europe*. Abstracts of the newborn screening experience and knowledge shared in both oral presentations and posters at the symposium are gathered here to assist in selecting presenters to attend virtually and posters to view online. Some abstract highlights include findings from pilot studies of new screening disorders, the value of screening older previously unscreened children, and benefits of second tier testing.

**Keywords:** newborn screening; ISNS; Europe



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## 1. Invited Presentations

### 100. Working Together in the Heart of Europe

Jim R Bonham

President, International Society for Neonatal Screening

The last year and a half has been a remarkable time for countries across the world. We moved from an era of normal social contact into the virtual age in one giant leap and sadly almost a quarter of a billion people have contracted COVID-19 with around 5 million tragically dying as a result.

Of course babies have continued to be born and I must pay tribute to the way in which newborn screening programs across the world continued to operate and to the dedication of the staff—all of you, who have made this possible—and the industries, including our sponsors, who have maintained supplies in this difficult time.

During the last 18 months we learned much about mass genetic testing in our populations and this has emphasized the potential for the technology to be applied in public health programs and newborn screening may be a beneficiary of this. We shall hear much more of this during our conference. The months of isolation also reminded us to value one another and the human touch, to make our societies grow and flourish.

In an exciting development on 28 June 2021 saw the first 'International Neonatal Screening Day' which ISNS has helped create, and we look forward to this developing in 2022.

Within Europe itself, we have seen a growing emphasis, supported by on-line meetings, to work with policy makers, MEPs, patient groups, the European Reference Networks and of course ISNS to help develop screening policy and practice. You will be hearing more about this during the meeting.

Ultimately; however, it is science and medicine that delivers life changing benefits for our children and their families and looking at the program we have much to learn and much to celebrate during the coming days of this exciting three day conference, I hope that you enjoy the talks and unlock some of the potential that they contain.

a confirmation test when using the cut-off value from this study. In addition, there was no significant difference in TSH levels in normal newborns with LBW and preterm newborns ( $p > 0.005$ ).

Conclusion: The reference value of TSH for normal newborns is 0.66–9.67 mU/L. Decreasing the referral value will increase the likelihood that an infant with transient congenital hypothyroidism will receive further treatment.

Keywords: Reference Value, TSH, Congenital Hypothyroidism

#### **P05. Incidence of Congenital Hypothyroidism in the Republic of North Macedonia in Correlation with TSH Cutoff Level**

Violeta Anastasovska, Milica Pesevska, Mirjana Kocova, Elena Sukarova-Angleovska, Nermina Fakovic and Senada Karishik

Department for Neonatal Screening, Faculty of Medicine, Ss. Cyril and Methodius University in Skopje, University Clinic for Pediatrics, Skopje, North Macedonia

The incidence of congenital hypothyroidism (CH) worldwide ranging from 1:2000 to 1:4000 newborns. Lower cutoff levels in screening programs have led to an increase in the proportion of detected cases with transient hypothyroidism, leading to increase of the overall incidence of primary CH.

Data from a newborn thyroid screening program over a 19-year period (2002–2020) were analyzed. Total of 354,422 (97.2%) neonates were screened for thyroid-stimulating hormone (TSH) level in dried blood spot specimens taken 48 h after birth, using DELFIA method. A TSH value of 15 mU/L was used as the cutoff point until 2010 and 10 mU/L thereafter.

Primary congenital hypothyroidism was detected in 202 newborns with overall incidence of 1/1755, and female to male ratio 1.04:1. Among neonates with primary CH, 144 (71.29%) had permanent CH with female predominance (female to male ratio 1.4:1), and 58 had transient CH (28.71%) with male predominance (female to male ratio 0.49:1). The lowering of the TSH cutoff level almost doubled the incidence of primary CH (1/1480) compared to the incidence in the previous period (1/2489). It is interesting that the incidence of transient CH in the period with lower TSH cutoff level (1/3751) was twelve times higher than the CH incidence detected until 2010 (1/45,625). Opposite, the incidences of permanent CH, before (1/2632) and after (1/2365) changing the TSH cutoff level, were slightly different.

Our findings showed that the lower TSH cutoff values have impacted the increased incidence of primary CH, especially of the transient CH, in the country.

#### **P06. Coverage with Neonatal Thyroid Screening in the Republic of North Macedonia, during 2002–2020**

Milica Pesevska, Violeta Anastasovska, Mirjana Kocova, Elena Sukarova and Nermina Fakovic  
University Clinic for Pediatrics, Department for Neonatal Screening, Skopje, North Macedonia

Neonatal thyroid screening is a reliable tool for early diagnosis and treatment of congenital hypothyroidism, the most common cause of preventable brain damage. A newborn thyroid screening program was established nationwide in 2007 in North Macedonia, after five years as a pilot study.

One screening center in the North Macedonia, located at the University Clinic for Pediatrics in Skopje, covers neonates born in 27 public and private birth centers all over the country. Thyroid-stimulating hormone (TSH) levels were analyzed from dry blood spots collected 48 h after birth on filter paper using the DELFIA method, during the period 2002–2020.

During this period, out of 364,574 live births 354,422 were screened. The coverage of the screened newborns was 97.2% on average, ranging from 91.1% in 2002 to 98.8% in 2020. There is continuously increasing rate of coverage in the last five years (98.3% in 2016, 98.2% in 2017, 98.3% in 2018, 98.6% in 2019 and 98.8% in 2020) as a result of improved education and training of the personnel in the births centers across the country. The biggest birth centers in the country, University Clinic of Gynecology and obstetrics (94.7%; 96.5%;



— 12<sup>th</sup> —  
**ISNS**  
— 2021 —

10 - 12 November 2021  
Luxembourg Going Virtual





# CERTIFICATE OF ATTENDANCE

This is to certify that

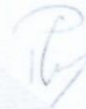
**Prof Violeta Anastasovska**

has attended the **12th ISNS European Regional Meeting, Luxembourg Going Virtual**, held online, from 10 - 12 November 2021.



Jim R. Bonham  
*ISNS President*

Kate Hall  
*ISNS 2021 LOC Chair*



Patricia Borde  
*ISNS 2021 President*