

NEWBORN SCREENING (NBS) IN SLOVAKIA – NEW APPROACH

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NEWBORN SCREENING CENTRE SK (NSC)
BANSKA BYSTRICA SK



2013 JOINT MEETING NBS>S and ISNS

ATLANTA May 5 - 10 2013



WHERE ARE WE FROM ?





NBS HISTORY IN SK

DIS.	START	TEST
CH	1985 2001	T_4 RIA \approx TSH _{RIA} TSH _{ILMA}
PKU	1978 1995	GUTHRIE Phe Fluoromet.
CAH	2003	17OHP _{ILMA}
CF	2009	IRT _{ILMA} IRT – Cl-

EXPANDED NBS SK – 1st. January 2013

SPECTRUM OF SCREENED DISORDERS NSC SK

PKU CH, CAH, CF

MSUD, GAI

IVA, MCAD, LCHAD, VLCAD, CPTI, CPTII, CACT,



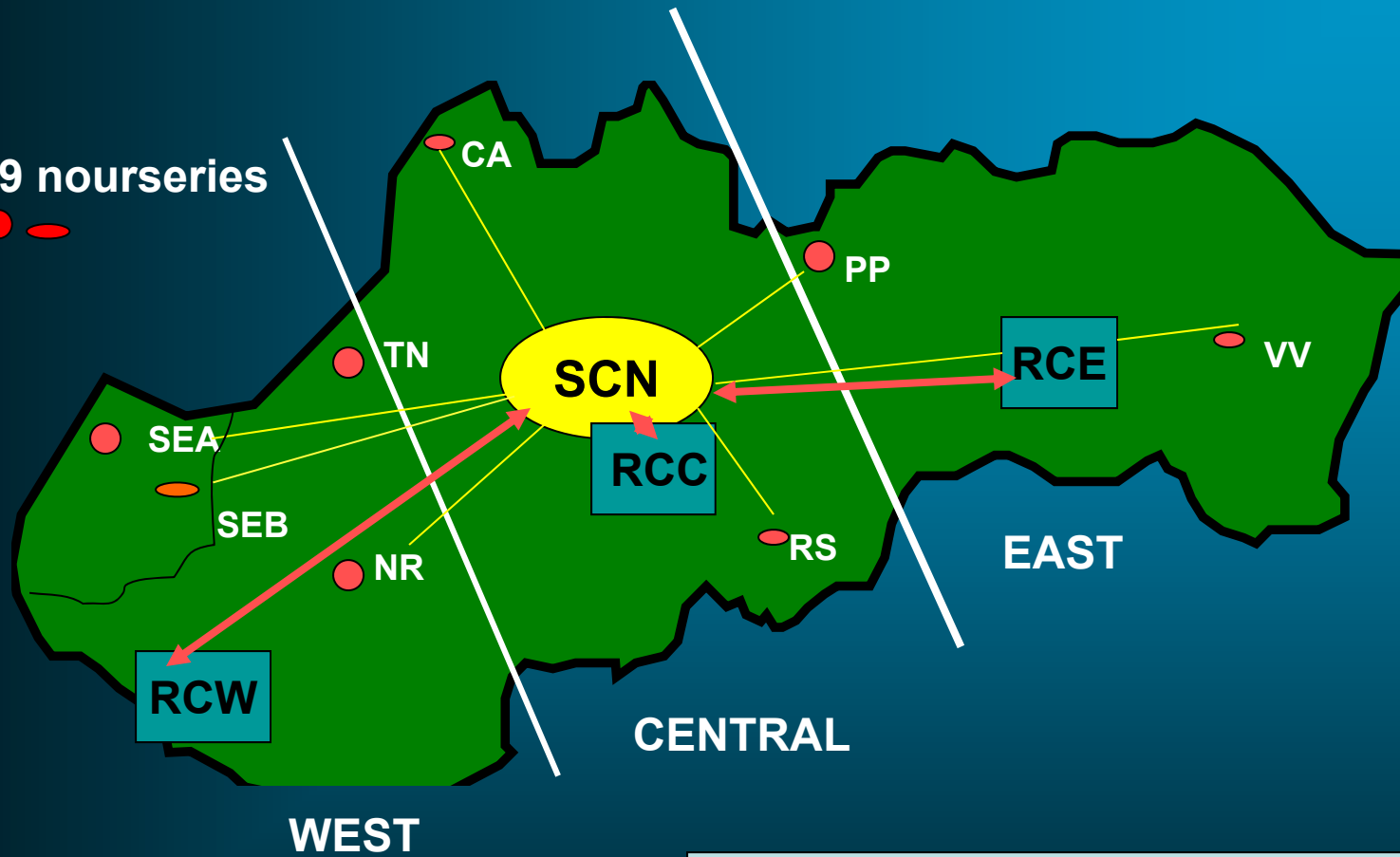
RED = ms/ms method





ORGANIZATION NBS SK

69 nurseries



RC = REGIONAL RECALL CTRs
RCW – West SK
RCC - Central SK
RCE - East SK



RESULTS OF NBS SK 1985 - 2012

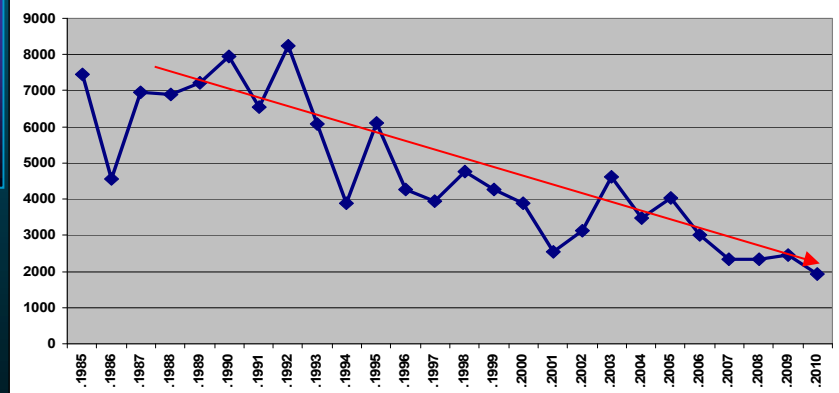
DISORDER SINCE No.SCREEN. No.POSITIVE SCR.PREVAL.

CH	1985	1,760 390	441	1 : 3 992
PKU	1995	983 109	166	1 : 5 922
CAH	2003	498 125	54	1 : 9 225
CF	2009	219 784	32	1 : 6 868

CH INCIDENCE

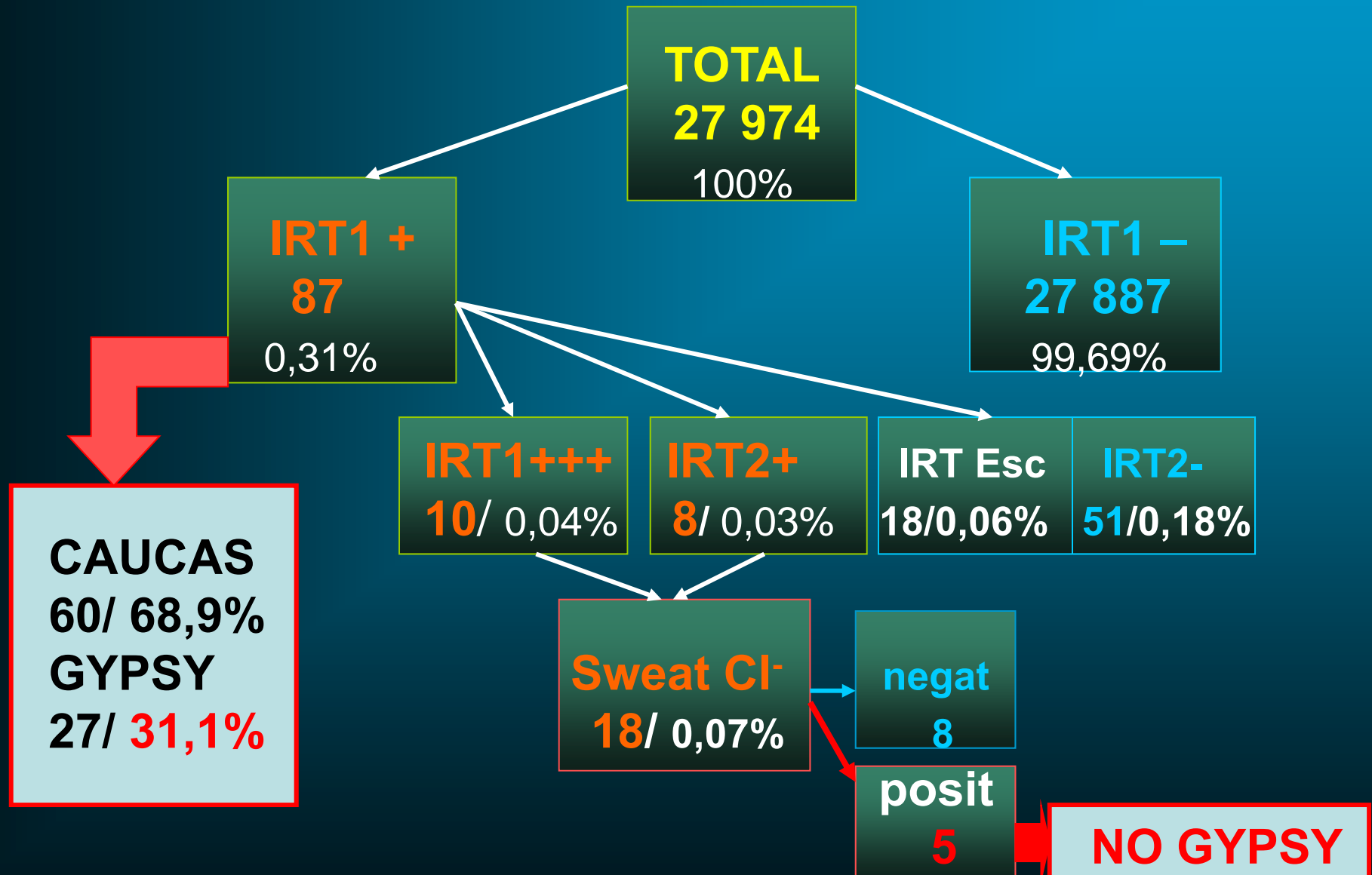
1 : 7 210

1 : 2 419



BEFORE INTRODUCTION
OF THE SCREENING :

SCF PILOT STUDY
May 5th – Oct 4th 2007



There was the

**real risk of false CF positivity in even 30%
newborns of Gypsy origin,**

In order

TO AVOID THE UNACCEPTABLE STRESS OF FAMILIES

and

INAPPROPRIATE MEDICAL & ECONOMIC BURDEN

**WE REALISED THE STUDY COMPARED OF NBS IRT1_{DBS}
IN NEWBORNS
OF CAUCASIAN AND GYPSY ETHNIC ORIGIN**

METHOD

PROSPECTIVE COMPARATIVE STUDY OF THE IRT_{DBS} LEVELS IN CAUCASIAN (MAJORITY) AND GYPSY (MINORITY)



DURING REGULAR NBS IN SLOVAKIA
1st MARCH – 31th MAY 2010



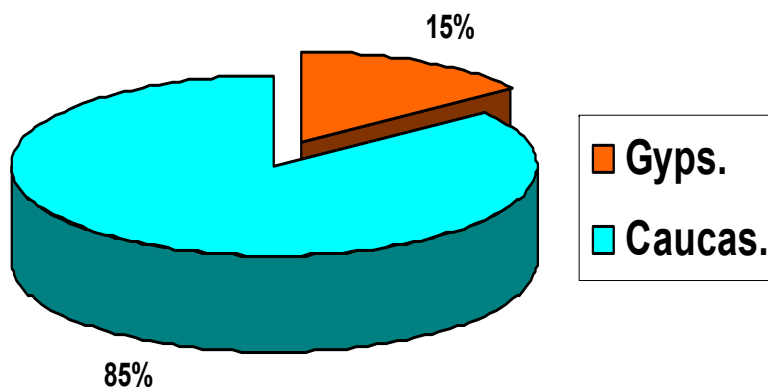
GYPSY PHENOTYPIC ETHNICITY WAS ASSIGNED
AS „R“ ON THE DBS SAMPLE CARD IN THE NURSERY



LABELING OF ETHNICITY WAS APPROVED BY ETHICAL
COMMITTEE OF THE HEALTH GOV.& NBSC SLOVAKIA
INDIVIDUAL DATA WERE ASSIGNED AS CONFIDENTIAL

Fig.3

THE NUMBER AND ETHNICITY IN THE CF STUDY 1st March – 31th May 2010



Caucasians	8 742
Gypsies	1 500
Total	10 242

Percentage of Gypsy Newborns - 15% is higher than percentage of the whole Gypsy population - 10%

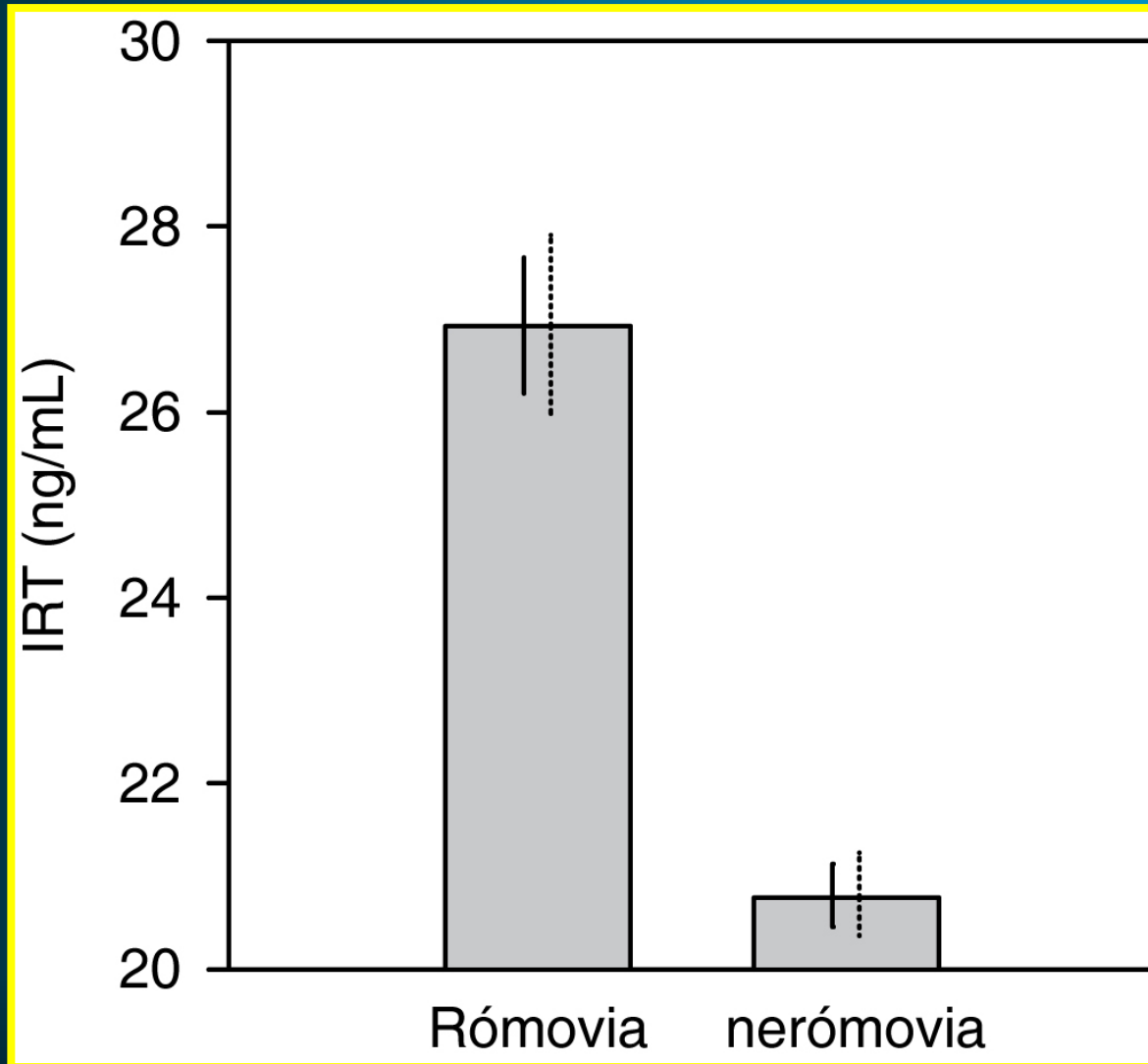
NATALITY RATE IN GYPSY POPULATION IS HIGHER THAN IN CAUCASIAN POPULATION

Mean Values and Intervals of the Confidence of IRT1 in Caucasian and Gypsy Newborns

	No. Newborns	Mean value	95% confidence interval		99% confidence interval	
Gypies	1500	26,92	26,20	27,66	25,98	27,90
Caucas.	8742	20,77	20,45	21,13	20,36	21,25

MEAN VALUES OF IRT1 ARE OF 23% HIGHER IN GYPSIES THAN IN CAUCASIANS

MEAN VALUES OF IRT1 IN CAUCASIANS AND GYPSIES
Bootstrap Mean Intervals of Confidence 95%= Solid Line, 99%= Dashed Line.



(t (10240) = 13.89, **p < 0.0001**).

FIRST CONCLUSIONS & TASKS

1./ TO **ADJUST THE CUT- OFF LIMIT** OF IRT1 AND IRT2 (ng/mL) IN GYPSY NEWBORNS



	CAUCASIAN	GYPSY
IRT1	72	84
IRT2	60	72

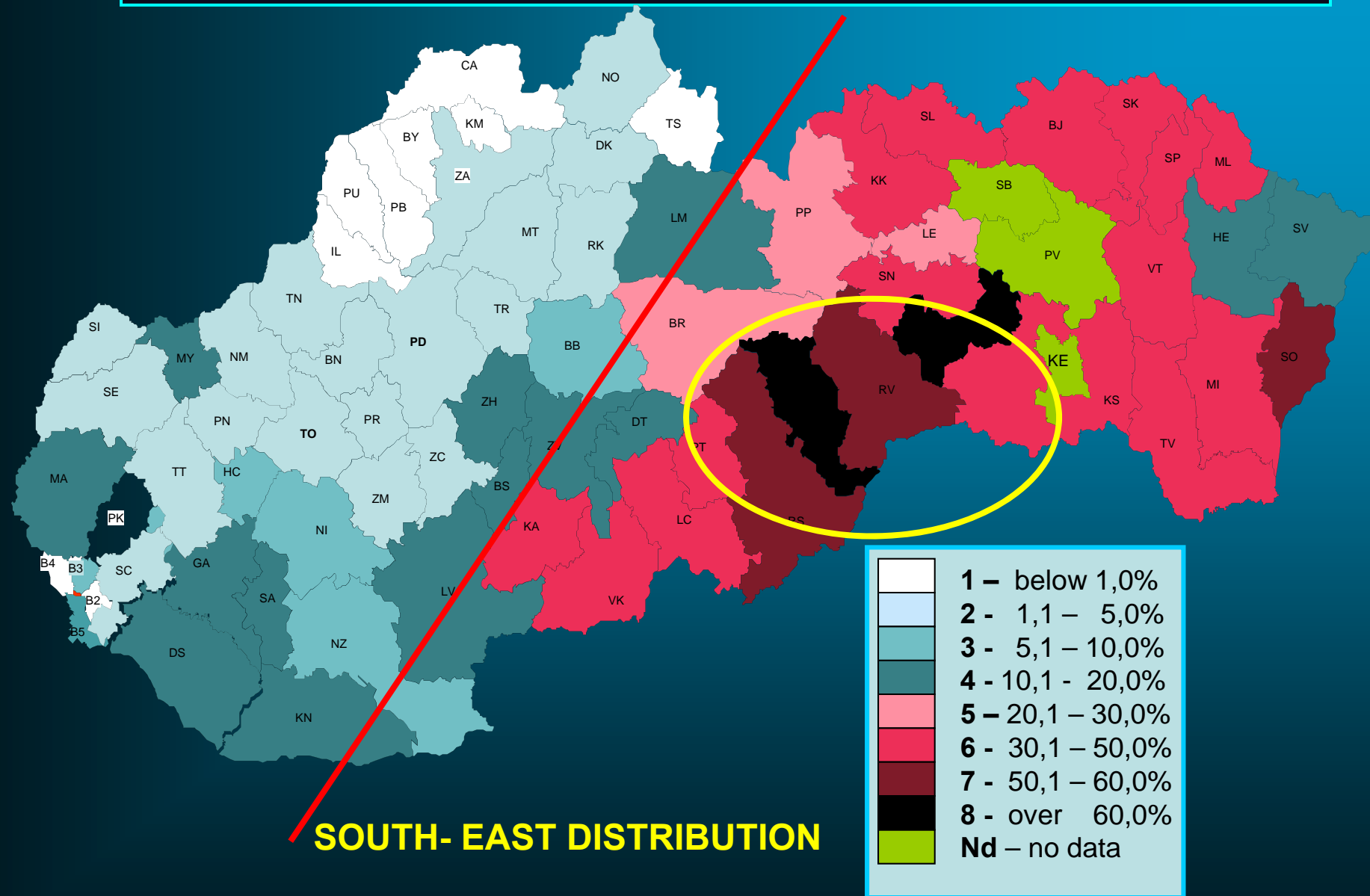
2./ **REGULAR LABELING OF THE ROMAN ETHNICITY** IN DBS SAMPLE - „R“

3./ **FOLLOW-UP OF THE GEOGRAPHIC DISTRIBUTION OF THE GYPSY NEWBORN POPULATION** – FOR PLANNING OF THE „ BURDEN OF HLTH SERVICE“

4./ **CONTINUOUS EVALUATION OF THE INCIDENCE OF CF IN GYPSY POPULATION**

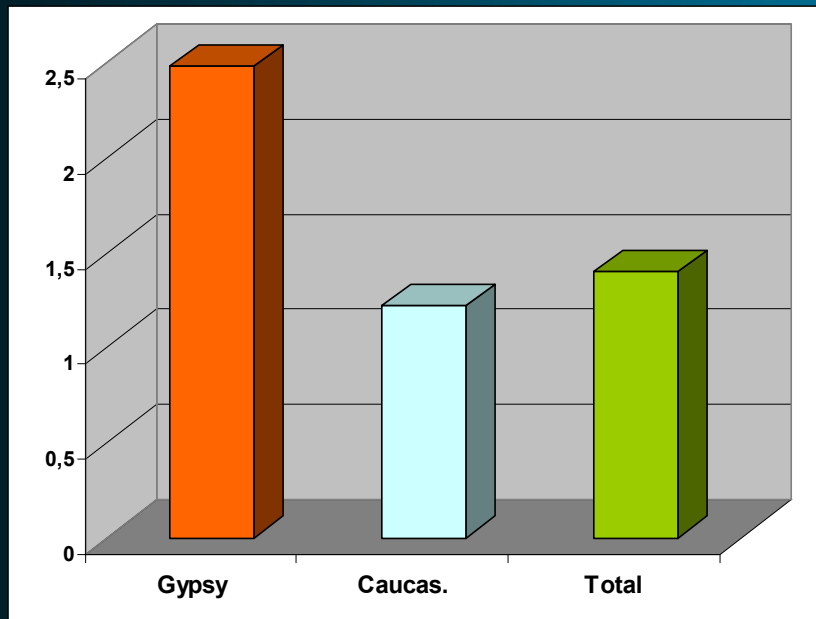
THE DISTRIBUTION OF THE GYPSY NATALITY IN SLOVAKIA

Percentage of Gypsy Newborns from Total Liveborn Infants - 2009



EVALUATION OF THE CF RECALL- RATE (RR) AFTER CHANGE OF GYPSY'S CUT-OFF LIMIT

1st March 2010 – 29th February 2011



Ethnicity	Total	RR
Gypsy	8428	209
Caucas.	51159	626
Total	59587	835

EVEN AFTER IMPROVING THE CUT- OFF LEVEL, GYPSY'S RR REMAINED TWOFOLD TO CAUCASIAN'S ONE

DETAILED RESULTS OF CYSTIC FIBROSIS SCREENING 2009 - 2012 YEARS

PARAMETER

VALUE / PERCENTAGE

NUMBER OF SCREEN. NEWBORNS	219 784
OF THEM GYPSIES (SINCE 2010)	21 115 / 9,6%
RR CAUCASIAN	0,58%
RR GYPSY	0,63%

NUMBER OF CONFIRMED CF	32
(ONLY CAUCASIANS)	
INCIDENCE OF CF	1 : 6 868

MEAN IRT1 CF	333,91 ng/mL
MEAN IRT2 CF	143,30 ng/mL
MEAN SWEAT Cl⁻ CF	94,47 mmol/L
GENOTYPE ΔF508del hom/ het/ other	13 / 12 / 6

MEAN TIME OF DEF. DIAGNOSIS	19,13 Day a.b.
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CONCLUSIONS

SLOVAK GYPSIES HAVE AN ELEVATED IRT LEVEL IN NBS, BUT CYSTIC FIBROSIS IS EXTREMELY RARE.

ELEVATED IRT LEVELS HAS BEEN DESCRIBED IN AFRO - AMERICAN INFANTS IN USA AND IN FRANCE IMMIGRANTS FROM NORTH AFRICA

ALTHOUGH A LOT OF STUDIES HAVE BEEN CONCERNING TO ETHNIC DIFFERENCES OF GENETIC MARKERS OF THE CF, WE HAVE FOUND NO STUDY PRESENTING THE ELEVATED IRT LEVEL IN GYPSY NEWBORNS

IN CONTRAST THIS, NO CASE OF ROMAN CF HAS BEEN DETECTED DURING NEWBORN CF SCREENING IN SLOVAKIA SINCE 2009. ONLY TWO CASES OF GYPSY CF ARE REGISTERED IN THE SLOVAK CF REGISTER (MORE THAN 30 YEARS OF EXISTENCE).

WE SEEN THIS FINDING AS A CONTRIBUTION TO THE KNOWLEDGE OF SPECTRUM OF THE GENETIC CHARACTERISTICS OF SLOVAK GYPSY POPULATION

Tatra Mountains – Nízke Tatry – Chopok – Slovakia nature





THANKS FOR YOUR KIND ATTENTION