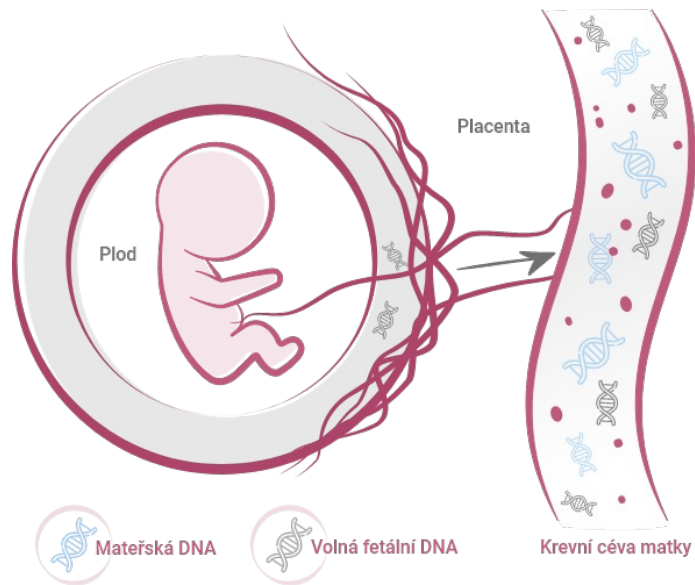
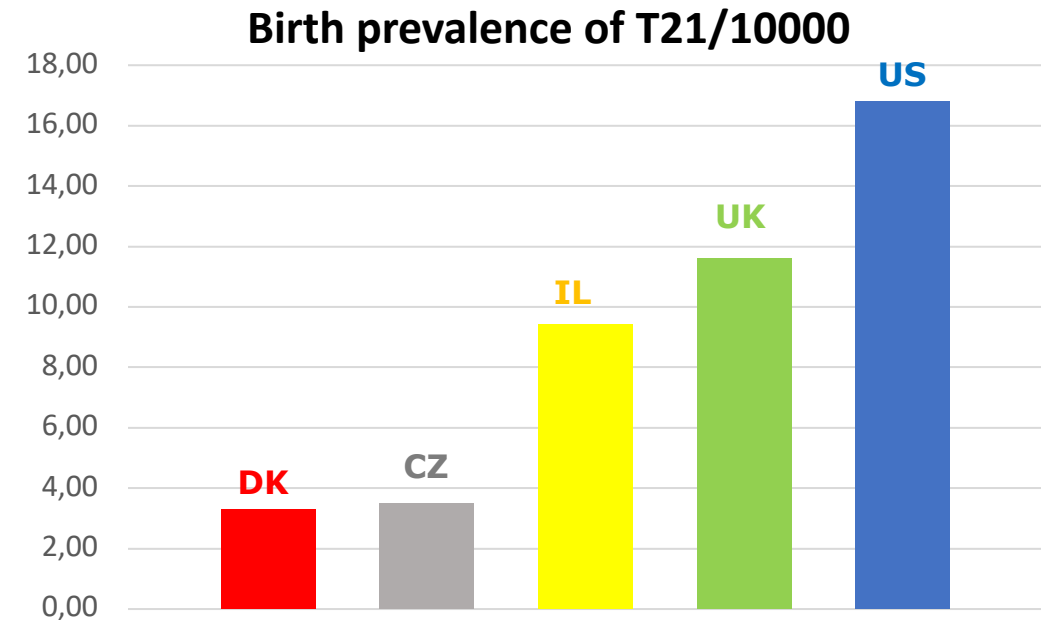


# NIPT in contingent screening protocol



# Arguments for systematic introduction of NIPT into antenatal screening

- **Expansion of diagnostic spectrum.**
- **Reduction the frequency of invasive tests.**



# NIPT in contingent screening

(recommendation SLG 2018)

HIGH RISK (INVASIVE) CONTINGENT

- COMBINED TEST RISK (CTR)  $>1/100$
- STRUCTURAL DEFECTS
- LOW PAPP-A/fbhCG

MIDDLE RISK (NIPT) CONTINGENT

CTR  $1/100 - 1/500$

LOW RISK CONTINGENT

---

FETAL ANOMALY SCAN 2nd TRIM

# **NIPT in contingent screening validation study**

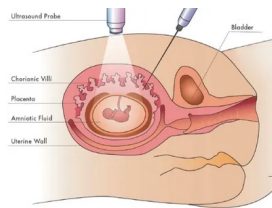
- **11378 naturally conceived low risk pregnancies**
- **Period Nov 2020 - Apr 2023.**
- **Average age 32yrs / 28.8% > 35**

# Screening protocol

## Combined Test



## INVASIVE



## LOW RISK

(*Integrated Test*)  
(*NIFTY by BGI*)  
(*Anxiety*)

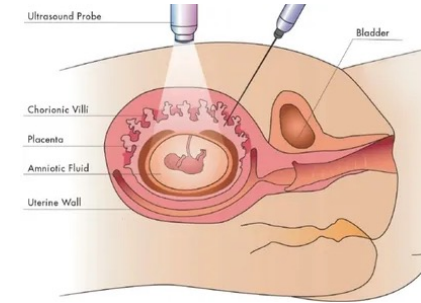
## NIPT



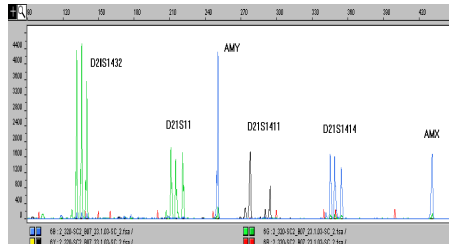
## II<sup>nd</sup> Trim Scan



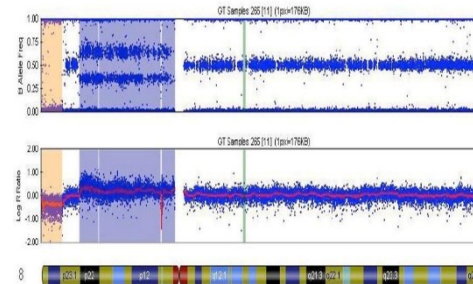
# Diagnostic procedures



## I. QF PCR



## II. CMA



## III. pES



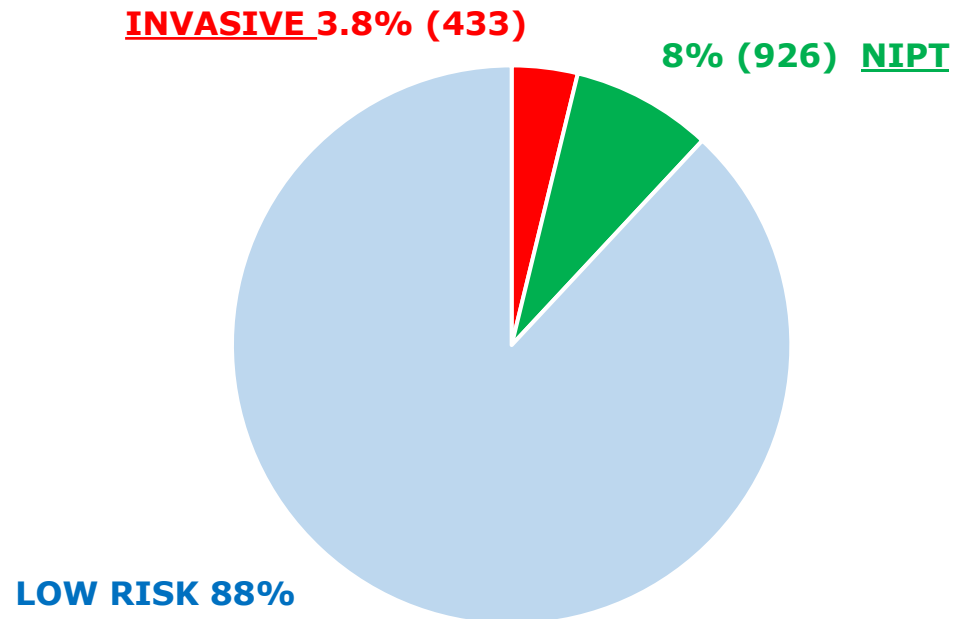
```
##fileformat=VCF4.2
##INFO=<ID=SVTYPE,Number=1,Type=String,
Description="Type of structure variant">
##INFO=<ID=END,Number=1,Type=Integer,
Description="End position of the variant described in this record">
#CHROM POS ID REF ALT QUAL FILTER INFO
```

```
1 160929435 rs7520618 G A . . SVTYPE=SNP;END=160929436
1 160932043 rs113387749 A . . SVTYPE=INS;END=160932043
1 160932206 rs5778188 C . . SVTYPE=DEL;END=160932207
1 160932771 rs2256505 A G . . SVTYPE=SNP;END=160932772
1 160934077 rs2481074 T A . . SVTYPE=SNP;END=160934078
1 160934818 rs1023115 A G . . SVTYPE=SNP;END=160934819
1 160935328 . AAA TGC . . SVTYPE=SUB;END=160935331
1 160935334 rs75452934 AA TC . . SVTYPE=SUB;END=160935336
```

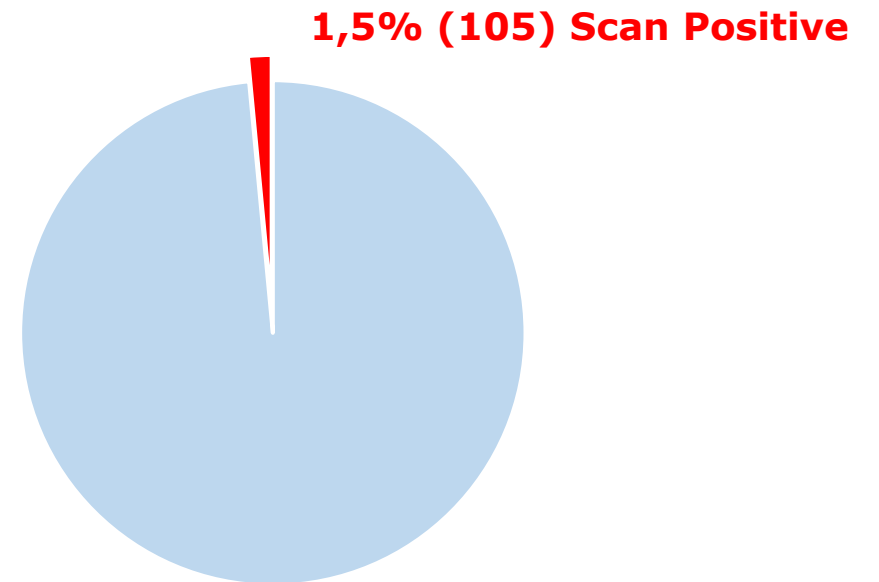
# Screening Results

11378 cases

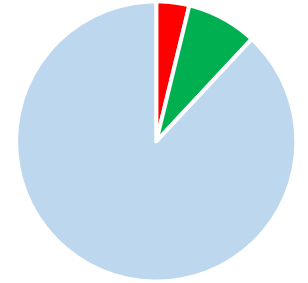
## First Trimester



## Second Trimester (66% cont.)

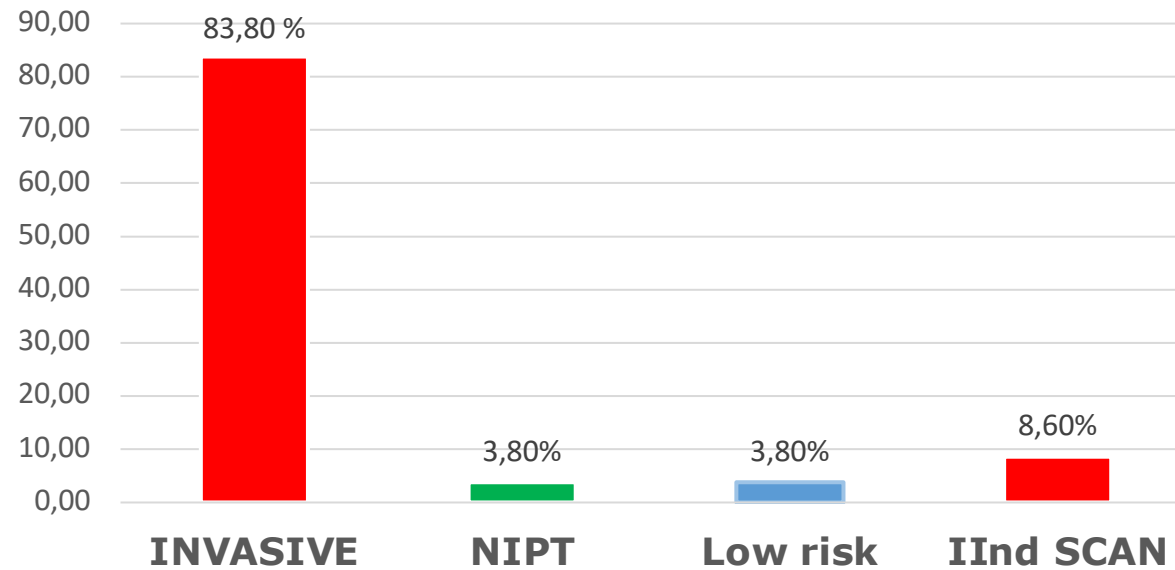


# RESULTS



## distribution of findings (105) by contingents

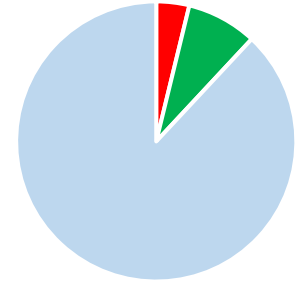
Dg. Procedures	Aneupl	CNV	Monogenic	Yield
653 (5.7%)	93	5	7	105 (15%)



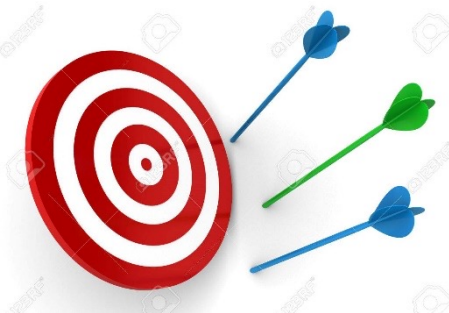


# NIPT RESULTS

NIPT contingent (656/926=70.8%)



No	Initial positives	True positives	Aneupl	CNV	NO_report	PPV
656	8 (1.2%)	2 (0.31%)	2 (+21)	0	4(0.6%)	25%



# CAN BE DIAGNOSED BY NIPT ?

(79%)

## ANEUPLOIDIES

Aneuploidy	T21	T18	T13	XO, XO mosaic	XXY	Triploidy	Total
NO	45	25	3	8	1	7	89
Replaceability By NIPT	100%	100%	100%	50%	100	0%	78 (87.6%)

## CNV

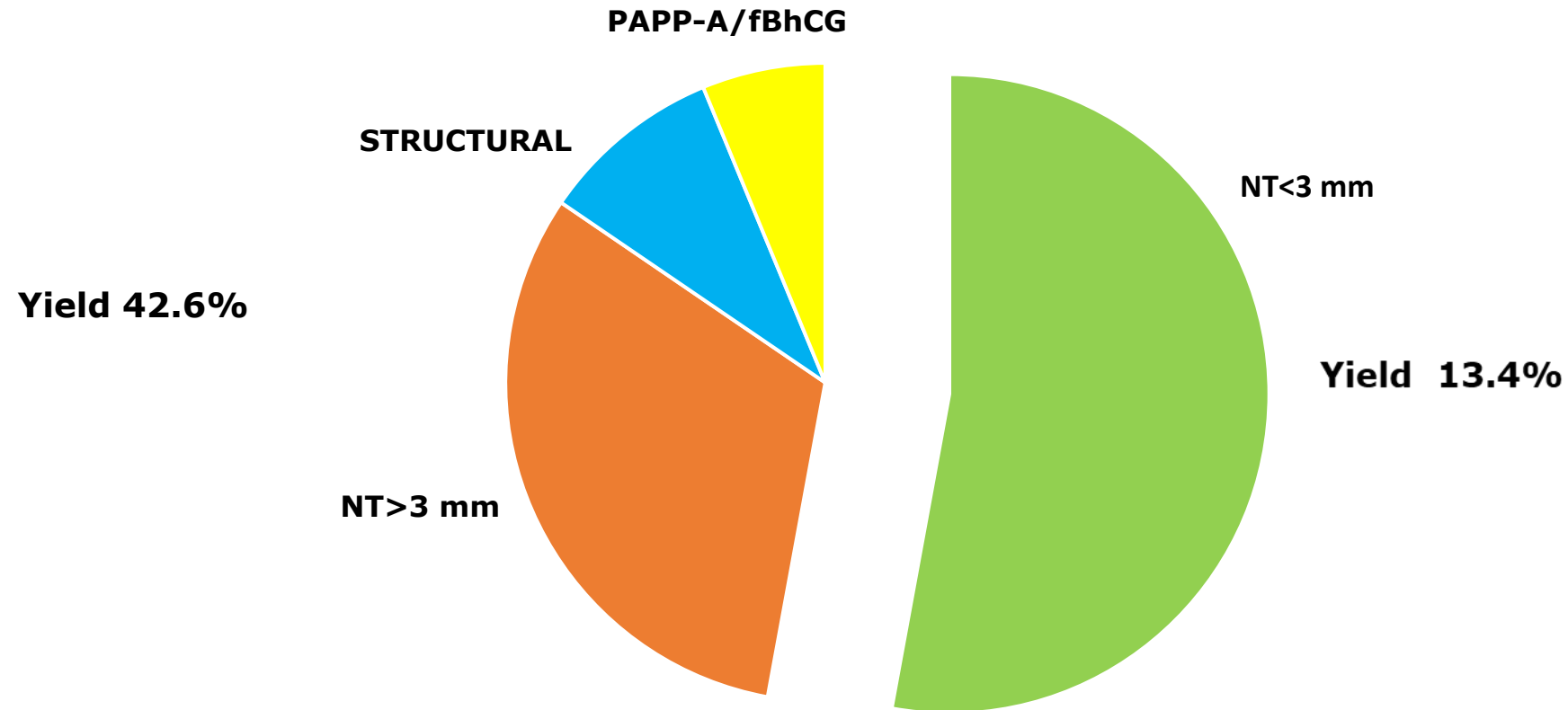
CNV	Length
5p15.33p14.2x1	23.8Mb
22q11.21x1	0.78Mb
22q11.21x1	0.74 Mb
3p26.3 x3	0.817 Mb
22q11.21x3	2.6Mb

## SNP

(CHD7)	(p.Asp1758Gly)
(GLI2):	(p.Leu1140Ter)
(SOS1):	(p.Lys170Glu)
(FGFR3):	(p.Ser249Cys)
(L1CAM):	(p.Glu309Lys)
(BICC1):	(p.Gly149Ser)
(TSC2):	(c.1947-2A>G)

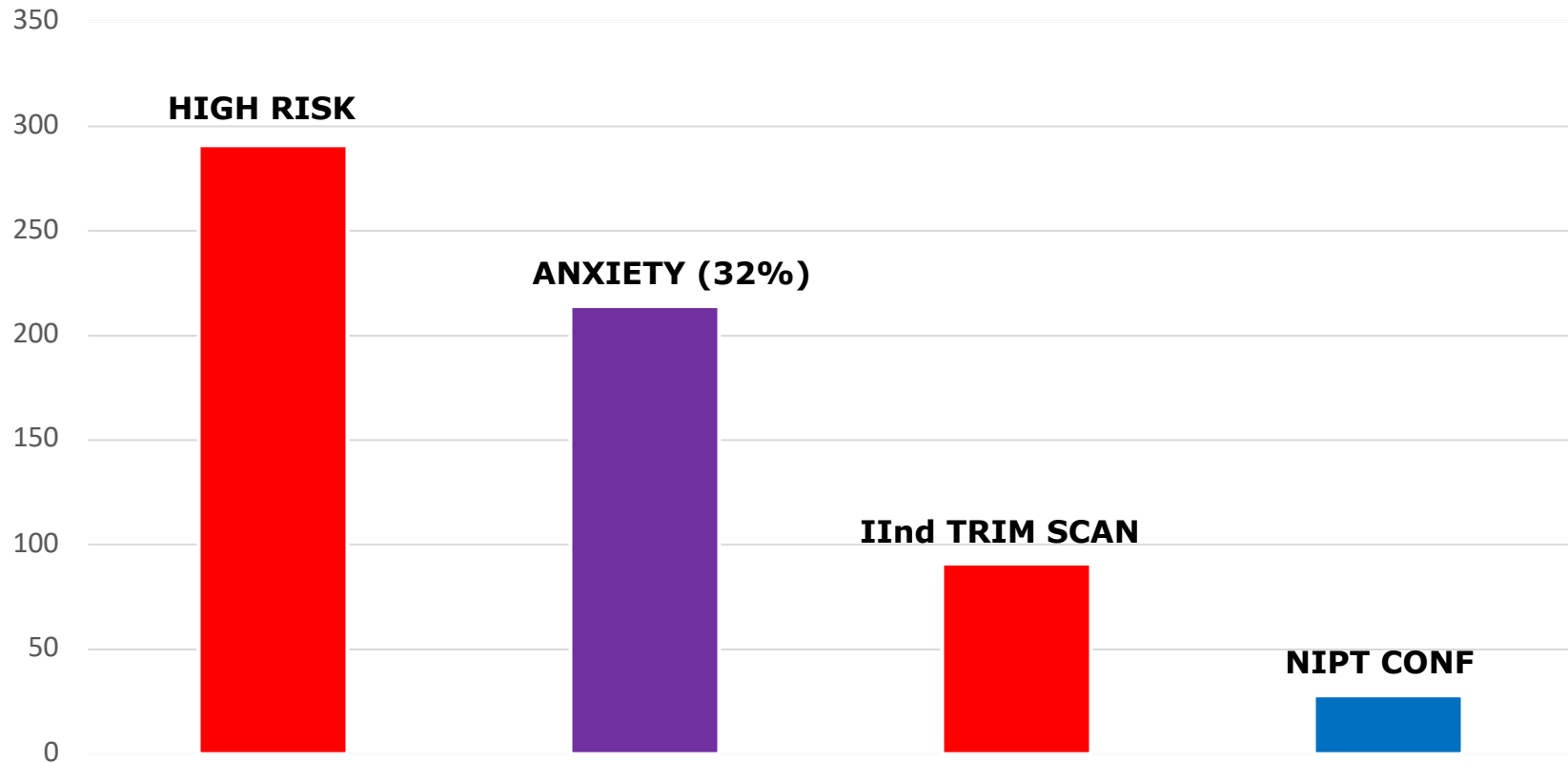
# INVASIVE contingent

(433/3.8%)



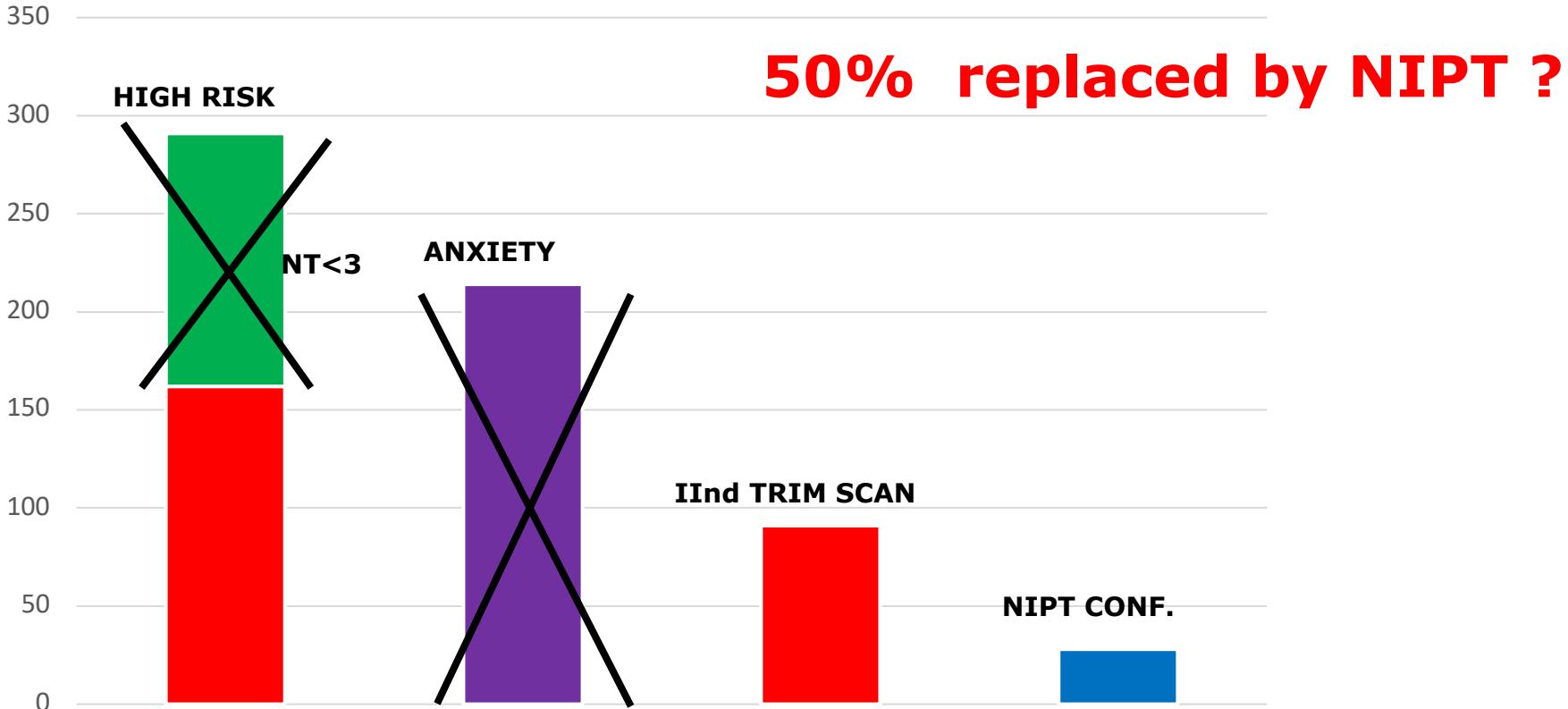
# Indications to diagnostic procedures

653/11378 (5.75%)



# Indication to diagnostic procedures

653/11378 (5.75%)



# **NIPT in contingent screening conclusion:**

- **NIPT contributed to 2% of aneuploidy (=T21) diagnoses**
- **The frequency of invasive procedures did not decrease**
- **Potencial of NIPT is in FTS positive and NT<3 mm**
- **NIPT could replace all invasive procedures indicated by maternal anxiety**
- **At least 3% of pregnancies are candidates for invasive procedure anyway**

# NIPT in contingent screening protocol



**\*1944**

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**Dagmar Smetanová**  
**Filip Zembol**  
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**Michael Němec**  
**Lenka Nguyen Thi Ngoc**



# Prague Spring Symposium

*7th International Symposium of Genetics and Reproductive Medicine*

The Centre of Medical Genetics and Reproductive Medicine, GENNET ([www.gennet.cz](http://www.gennet.cz)), a member of FutureLife group is delighted to announce that The Prague Spring Symposium 2024 will be held in Prague on **Friday, May 24th, 2024.**

*Registration:*  
[www.praguespringsymposium.com](http://www.praguespringsymposium.com)

## *Venue:*

Prague Marriott Hotel, Praha 1, V Celnici 8

## *Expert guarantor of the event:*

Dr. David Stejskal