Contingent Model in Switzerland

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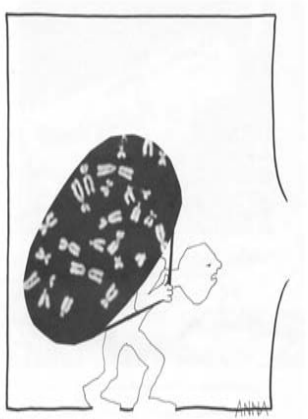
NIPT



"ANNA"
Schweizerische Ärztezeitung, 1999;80:16

New questions

When: Timing of NIPT

How : Implementing NIPT into the existing screening concept

Costs: Cost-benefit calculations

Law on compulsary health insuareance in Switzerland

Legal Regulations (KVG/KLV)

- Ultrasound screening at 11-14 and 20-22 weeks
  or if indicated in pregnancies at risk
- CVS/AC in pregnancies with increased maternal age (≥35 yrs) or a comperable risk (cut off: 1:380)
- Combined Test was not officially listed but the costs were accepted
- Combined Test now reimbursed (since 2015)
- NIPT now reimbursed (since 7/2015)

Implementation procedure of NIPT

- **Two companies applied for reimbursement at Swiss Federal Office of Health (BAG)**
- **BAG invited the professional organisations**
- **BAG asked for a proposal how NIPT can be implemented in routine antenatal care**
- **This is the formal procedure to include an new lab-test in the list of analysis**

NIPT and professional societies

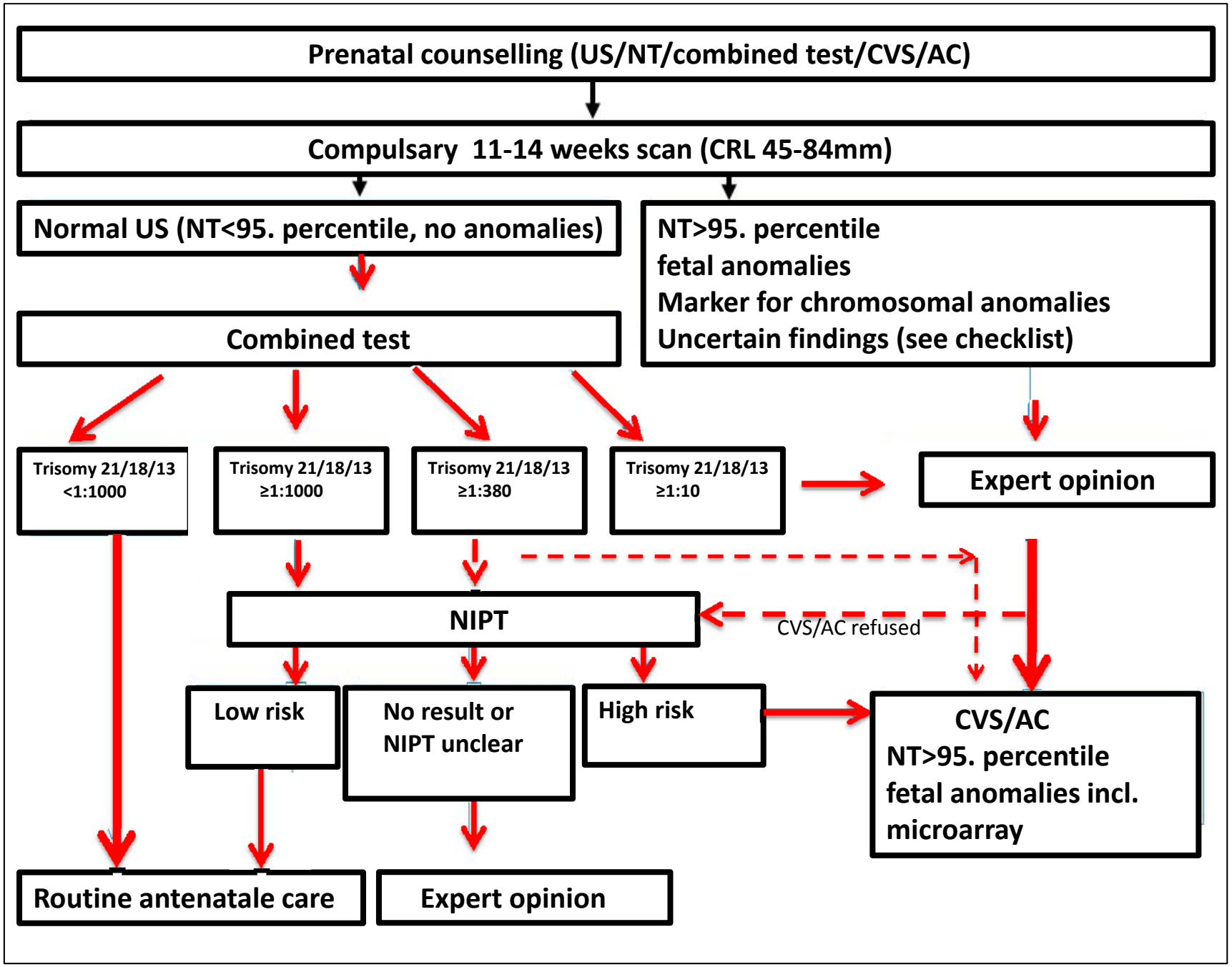
- Swiss society of Ob/Gyn
- Academy for Feto-Maternal Medicine
- Swiss Society for Ultrasound in Medicine („pregnancy-commission“)
- Swiss Society of Medical Genetics
- Swiss Study Group-1.st Trimester Testing
  („voluntary“ co-operation between laboratories and SGUM)

Arguments in favour of NIPT

- **Superior test performance as compared to combined test**
- **Drastic reduction of invasive testing**
- **Ethical concerns (screening for trisomy 21 in a national program vs. to cover invasive testing /combined test but withhold NIPT)**

Consensus of all societies involved

- Non-invasive screening should be offered for all pregnancies if 11-14 scan is normal
- NIPT should not be considered as a replacement for ultrasound
- Invasive options should be maintained


Prenatal counselling (US/NT/combined test/CVS/AC)
Compulsary 11-14 weeks scan (CRL 45-84mm)
Normal US (NT<95. percentile, no anomalies)
NT>95. percentile
fetal anomalies
Marker for chromosomal anomalies
Uncertain findings (see checklist)
Combined test
Trisomy 21/18/13 <1:1000
Trisomy 21/18/13 ≥1:1000
Trisomy 21/18/13 ≥1:380
Trisomy 21/18/13 ≥1:10
Expert opinion
NIPT
CVS/AC refused
Low risk
No result or NIPT unclear
High risk
CVS/AC
NT>95. percentile
fetal anomalies incl. microarray
Routine antenatale care
Expert opinion

Current regulation for NIPT

- Contingent screening strategy including first trimester combined screening and NIPT (NIPT after combined test)

- Reimbursement in cases with intermediate risk for trisomies 21, 18, 13 ≥ 1:1000

Requirements to request NIPT

- **Combined test including ultrasound first**

- **Certification for 11-14 weeks scan (appropriately trained sonographers with annual audit, n=1088)**

- **Use of accepted software for risk calculation (FMF Germany or FMF London)**

Consensus group
- 17 Laboratories
- Representatives of SGUM/SGUMGG, AFMM, Swiss Society of Medical Genetics

Certain degree of standardisation among the laboratories

- **Fetal fraction rate**
- **Standard forms**
- **Data collection**

CH-1TT-NIPT (organization)

Members	
Laboratories:	FAMH medical genetics
Medical doctors:	FMH Gynäkologie und Geburtshilfe mit Fähigkeitsausweis Schwangerschaftsultraschall der SGUM FMH Medizinische Genetik FMH Gynäkologie und Geburtshilfe mit Schwerpunkt fetomaternale Medizin
Steering group (NIPT):	Board U. Wiedemann Laboratories M. Krüger (co-chair), F. Bena, C. Noppen, R. Spiegel, M. Morris (co-chair) SGMG I. Filges SGUM/SGUMGG S. Tercanli SGGG/AFMM L. Raio

Good News

NIPT 15.7.2015-14.7.2016

Pregnancies per year in Switzerland: n= 86.559

First-Trimester-Testing:

17 certified (CH-1TT) and 2 not certified laboratories offering 1.-TT in CH

1.-TT (tests/year): n= 65.500 (76%)
(data from all certified laboratories)

1.-TTs with risk (T 21) > 1/1000: ~ 14.0%
(data from 11 certified laboratories)

Good News

NIPT 15.7.2015-14.7.2016

Expected rate of NIPT`s ca. 14-15% (14.000)

risk>1:1000	risk<1:1000	unknown
35%	60%	5%

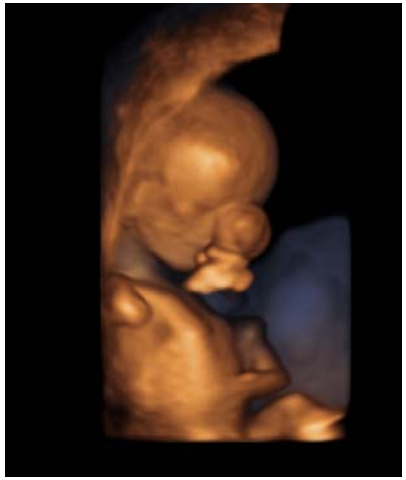
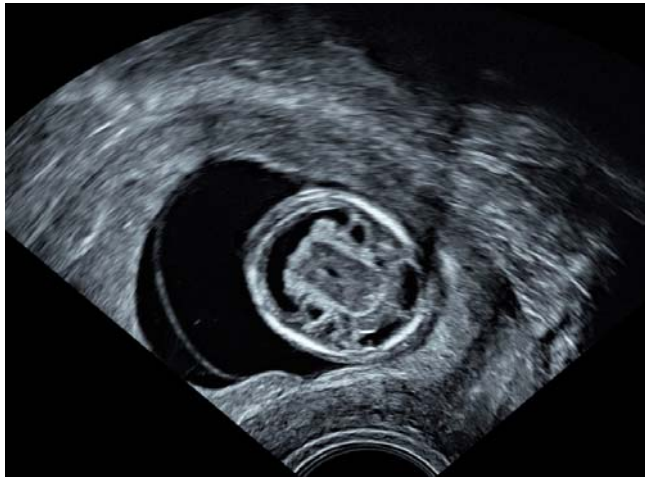
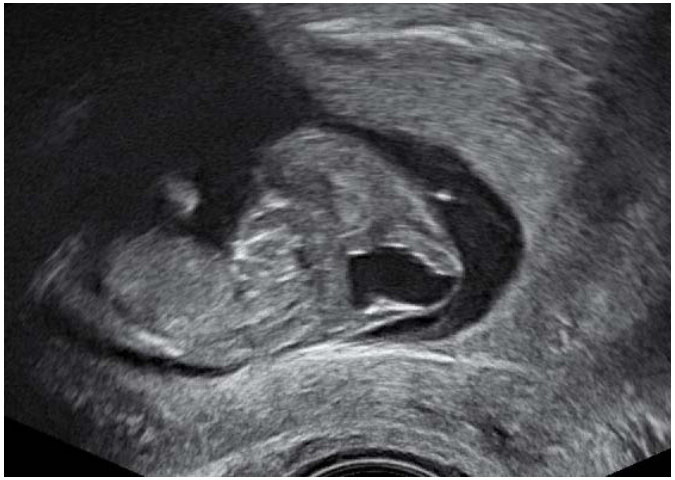
NIPT normal	NIPT abnormal	unclear/no result
96%	1.7%	2.3%

False positive cases: n= 21 (4 trisomy 21, 7 trisomy 18, 10 trisomy 13)
False negative cases: n= 2 (1 trisomy21, 1 trisomy13)

Bad News

Misuse of NIPT

- Focussing on NIPT may result in higher rates of misdiagnosis of other anomalies /chrom. defects
- Increasing rate of late diagnosis
- Increasing rate of late terminations

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Noninvasive prenatal testing: more caution in counseling is needed in high risk pregnancies with ultrasound abnormalities

Beatrice Oneda [a,*], Katharina Steindl [a], Rahim Masood [a], Irina Reshetnikova [a], Pavel Krejci [a], Rosa Baldinger [a], Regina Reissmann [a], Malgorzata Taralczak [a], Adriano Guetg [a], Josef Wisser [b], Jean-Claude Fauchère [c], Anita Rauch [a]

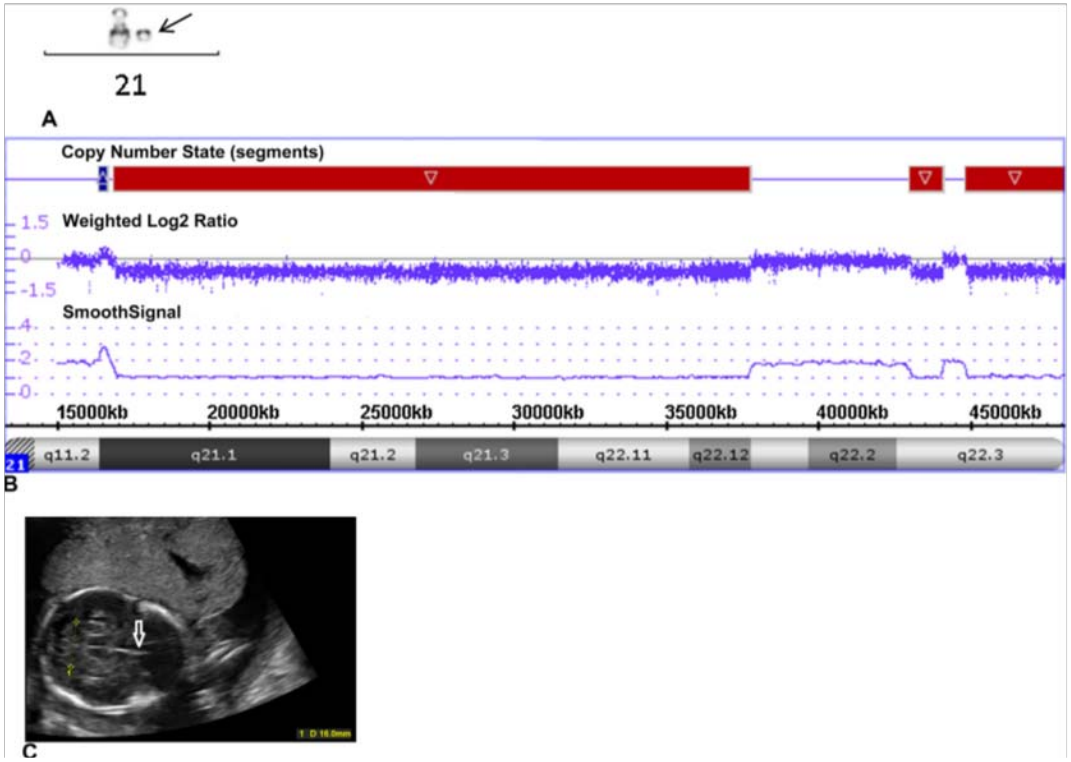

Fig. 1. Case 1: (A) GTG-banding showing complex rearrangement of one chromosome 21 (indicated by an arrow). (B) Example of chromoanasynthesis. Results of the CMA showing the duplications and deletions observed in the amniocytes: it consisted of one duplicated segment of 255 kb, and three deleted segments of 20.8 Mb, 1.06 Mb and

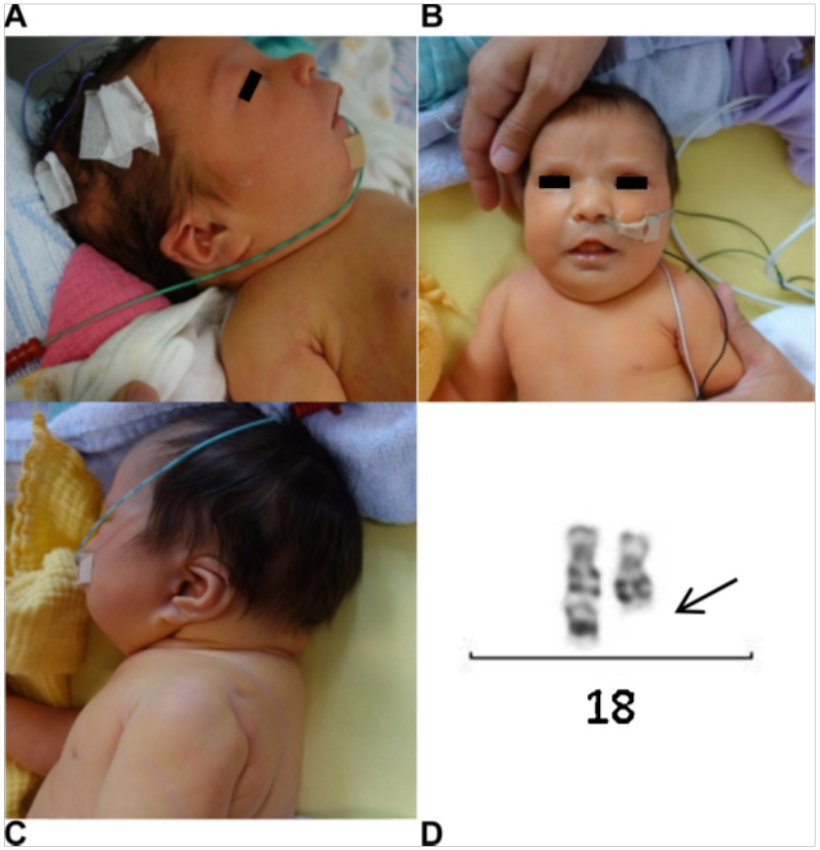

Fig. 2. (A–C)Photographs of case 3 at day 2 after birth. Note facial dysmorphism with up-slanting narrow palpebral fissures, hypertelorism, flat nasal root, flat long philtrum, thin and tented upper lip, abnormal helices, micrognathia and short neck with skin folds. (D) GTG-banding showing the partial monosomy 18q (indicated by an arrow).

In summary, we suggest more caution and education for patients and counselors and recommend a more accurate consideration of the testing indications