UNIKLINIK RWTH AACHEN
Institut für Geschichte, Theorie und Ethik der Medizin

Disability Detection and Foetal Decision Making through NIPT

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University of Sussex, 16.06.2014

Outline

1. Prenatal testing in Germany
2. NIPT in Germany
3. Regulatory Aspects
4. Ethical Aspects

1. Prenatal testing in Germany

- Implementation of invasive prenatal testing through a research program 1973-1978
- since 1975 covered by national health insurance >> immediate increase in utilisation
- Infrastructure of the German health care system in prenatal testing now characterized by:
  - administration on a federal state - level
  - many physicians in private practice
  - deficits (quantitative and qualitative) in accompanying genetic counselling processes
  - interprofessional conflicts of interest

Indikationen für die invasive PND

Indikationen	Anteil an durchgeführter invasiver PND
Erhöhtes mütterliches Alter	71,4 % (1022/1431)
Auffälliger mütterlicher Serumbefund (Triple-Test)	11,6 % (166/1431)
Psychische Indikation	8,3 % (119/1431)
Auffälliger Ultraschallbefund	0,8 % (12/1431)
Hohes Risiko (≥ 25 %) für monogen bedingte Erkrankung	3,0 % (43/1431)
Eltern Träger einer balancierten Chromosomenstörung	1,3 % (18/1431)
Vorheriges Kind mit Chromosomenstörung	3,6 % (51/1431)

(zusammengestellt aus: Nippert 2001)

Pränataldiagnostik von 1996-1999 in der Bundesrepublik Deutschland

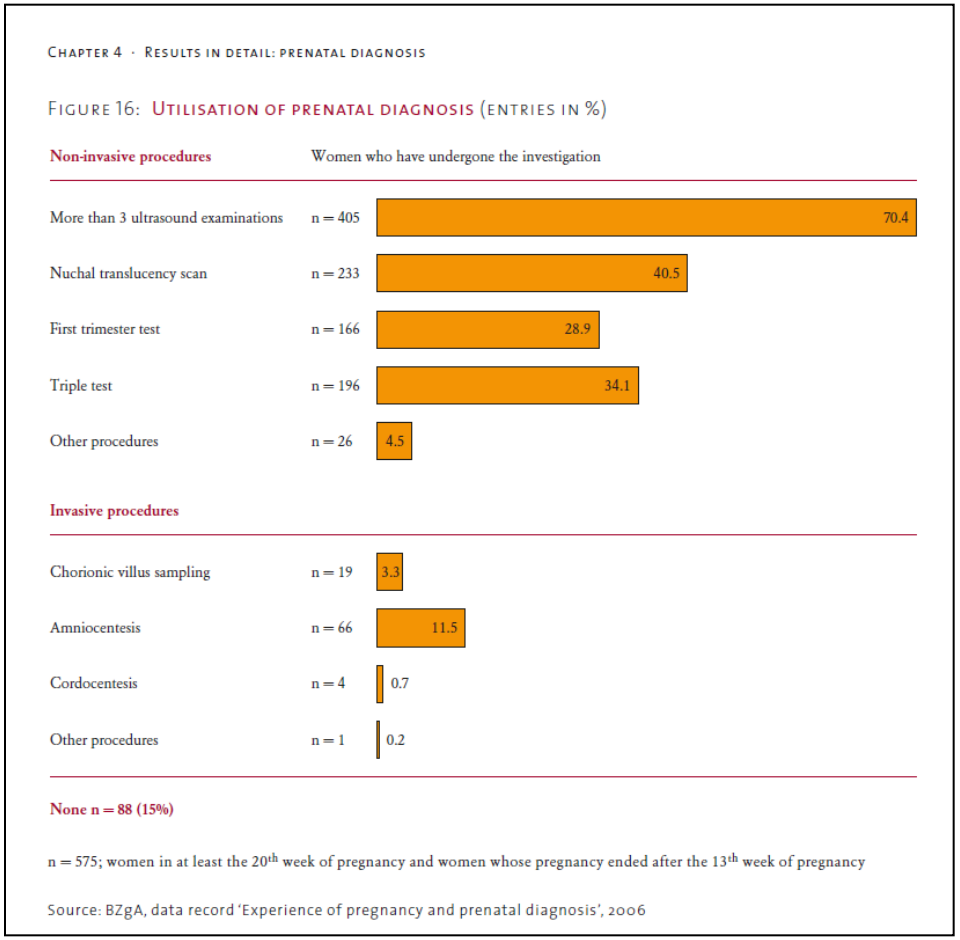
	1996	1997	1998	1999
Lebendgeburten[I]	796.013	812.173	785.034	770.744
Chorionzottenbiopsie (gesamt)	4.145	4.558	4.539	4.310
Alte Bundesländer	3.891	4.371	4.386	
Neue Bundesländer	254	187	153	
Amniozentese (gesamt)	58.186	62.667	62.419	63.010
Alte Bundesländer	54.439	58.250	58.111	
Neue Bundesländer	3.747	4.417	4.308	
Amniozentese + Chorionzottenbiopsie (gesamt)	62.331	67.225	66.958	67.320
Fehlgeburten als Komplikation nach PND[II]	ca. 600	ca. 700	ca. 700	ca. 700

Die Zahl der Untersuchungen wird in den neuen Bundesländern erst seit 1996 systematisch durch die Kassenärztliche Bundesvereinigung (KBV) erfasst. Zur Situation im früheren Bundesgebiet vor 1996 vgl. Tabelle VI im Anhang.
i) Statistisches Bundesamt 2002
ii) Annahme: 1 % Abortrisiko (vgl. Bundesärztekammer 1998)

National Ethics Council (2003) Genetic testing before and during pregnancy. Opinion. p. 51.

1. Prenatal testing in Germany

- More than 85% of all pregnant women have at least one additional prenatal testing procedure
- Aims of prenatal testing:
  - Ensuring baby‘s health: 61.6%
  - An aid to deciding wether to terminate the pregnancy in the event of a disability: 44.0%
  - Part of general prenatal care: 36.8%
  - My doctor wanted me to do so: 25.6%
  - My doctor had (very) strong influence on my decision for or against prenatal diagnosis: 52%



Results of a representative survey in Germany of the Federal Centre for Health Education (BZgA) in 2003

2. NIPT in Germany

- Close to 10 000 PraenaTests® (Lifecodexx) in Europe (focus on Germany, Switzerland, Austria) since launch (August 2012)
- 2013: 30 000 NIPT-tests in Europe (5000 in Germany)
- NIPT in 3% of all high risk pregnancies (maternal age or fts)
- In comparison: NIPT in 50% of all high risk pregnancies in the U.S.

Lutz, Michael (CEO LifeCodexx) „PraenaTest® Setting Standards in the Field of Non-Invasive Prenatal Testing (NIPT) for Common Fetal Aneuploidies“, Prenatal Molecular Diagnostics Europe (Conference), 13.5.2014, Lisbon

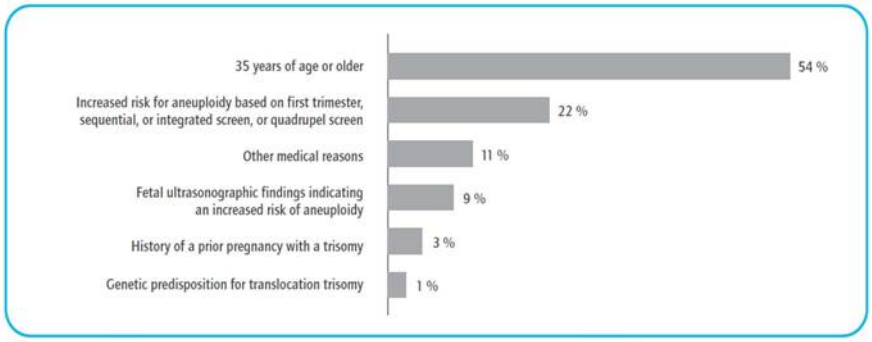


Figure 3: Summary of clinical indications for application of PraenaTest®

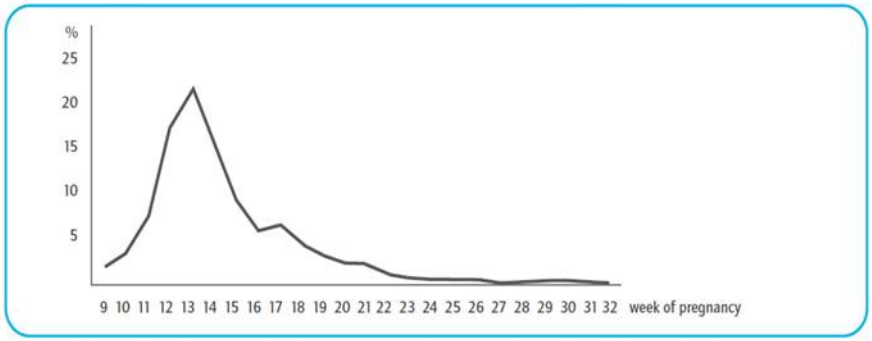


Figure 4: Overview about week of pregnancy of women who ordered PraenaTest®

- Approximately 148,500 MaterniT21 PLUS (Sequenom) tests in 2013 in the U.S.; 143% more than in 2012; 93% of accessions from U.S. patients [Sequenom, Financial results report 2013]

Schöck U et al., Non-invasive prenatal testing (NIPT): Laboratory experiences of PraenaTest®. Annual Meeting of the German Society of Human Genetics; Essen 2014

3. Regulatory Aspects

Professional guidelines

- Consensus statement (NIPT D, A, CH; 2013)
  - Only in high-risk pregnancies
  - After 11+0 weeks of gestation
  - Only in combination with high-resolution ultrasound
- German Medical Association (BÄK)
  - Guidelines on prenatal testing (1998)
- German Society of Human Genetics
  - Statement on prenatal diagnosis and termination of pregnancy (1993)
  - Statement on the analysis of fetal DNA in the maternal blood (11/2012)

Law

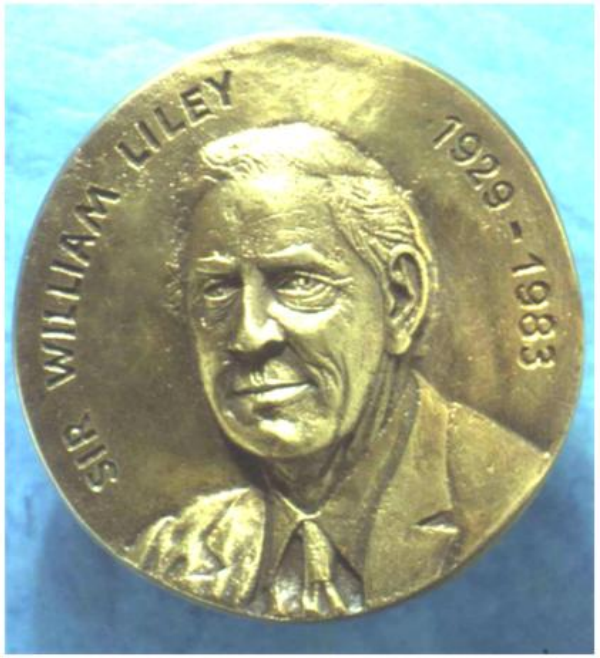
- German Genetic Diagnosis Act (2009)
  - §15 Prenatal genetic testing
  - Genetic Diagnostic Commission (GEKO)
    - Statement on NIPT (4/2014)
- Law on Conflicts in Pregnancy (2009)
  - §2a Disclosure and counselling in special cases

4. Ethical Aspects

The Fetus as a Patient?

- More personal information of the fetus will be available.
- More predictive genetic information will be available.
- More therapeutically relevant genetic information will be available.

- The question of the right not to know of the future child will be raised with heightened tension.
- The limitations of the principle of reproductive autonomy as the leading ethical principle in prenatal testing will be more obvious.



The William Liley Medal of the International Society of the Fetus as a Patient

Thank you for your attention