Epidemiology and Ultrasound screening of NTD in France.

Romain Favre
CMCO-HUS
Strasbourg France

Plan

- Recent published epidemiological data in France
- Analysis of the activity in our Fetal Medicine center of CMCO in Strasbourg
- Influence of the loss of the MSS of Down syndrome in the second trimester (α fœto-protein)
- Evolution of the prenatal Screening strategy

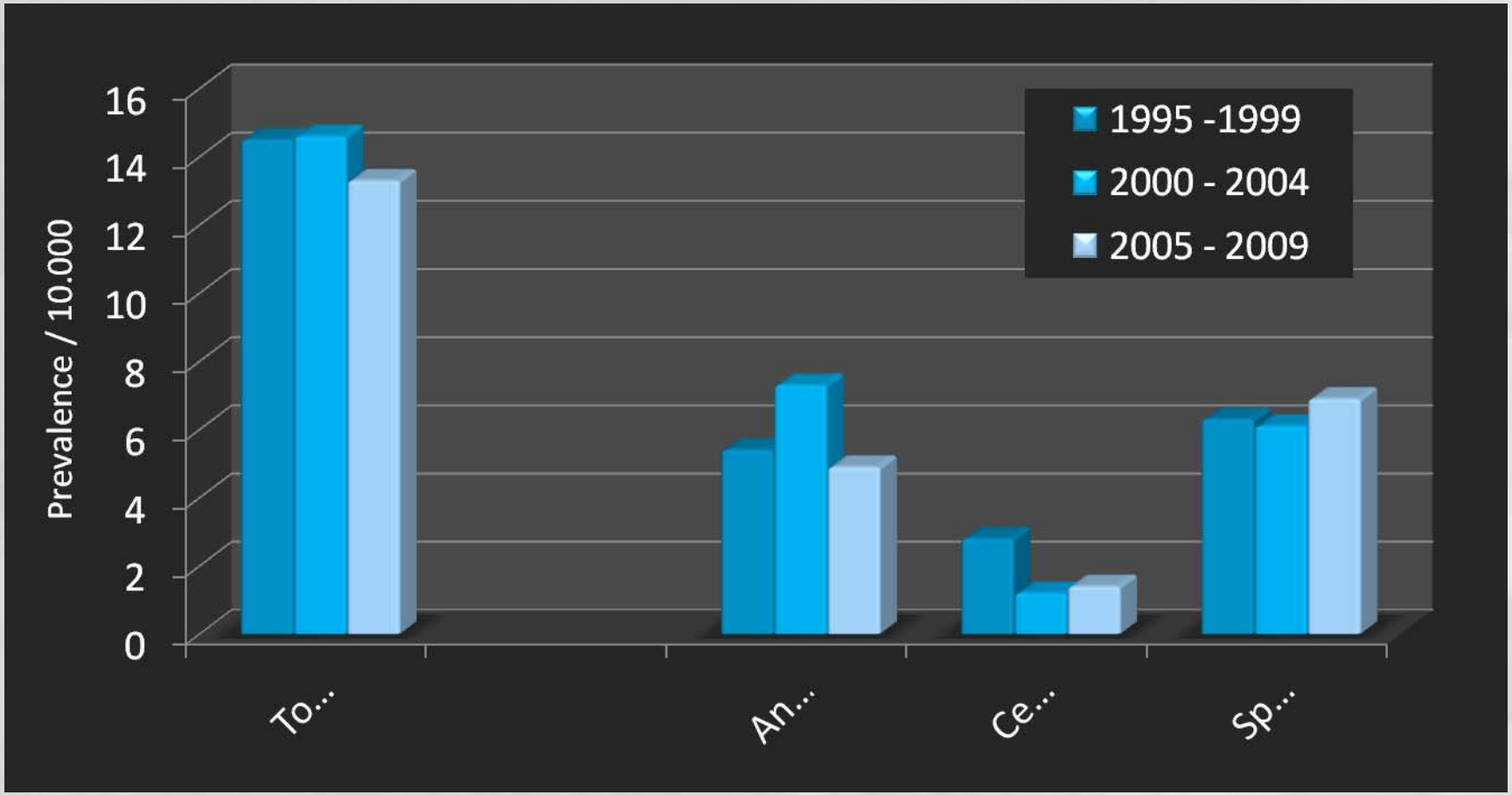
Epidemiological datas

- **Retrospective study from data of the Registry of Congenital Malformations of Alsace between 1995 and 2009.**
- **23.000 births per year, Total of 192.321 births**
- **272 cases of NTD – Prevalence 12/10.000**
  - **113 Anencephaly (41 %)**
  - **35 Cephaloceles (13 %)**
  - **124 Spina bifida (46 %)**

D. Timbolschi, R. Favre, B. Doray. Fetal Diagn. Ther. 2014

Epidemiological datas

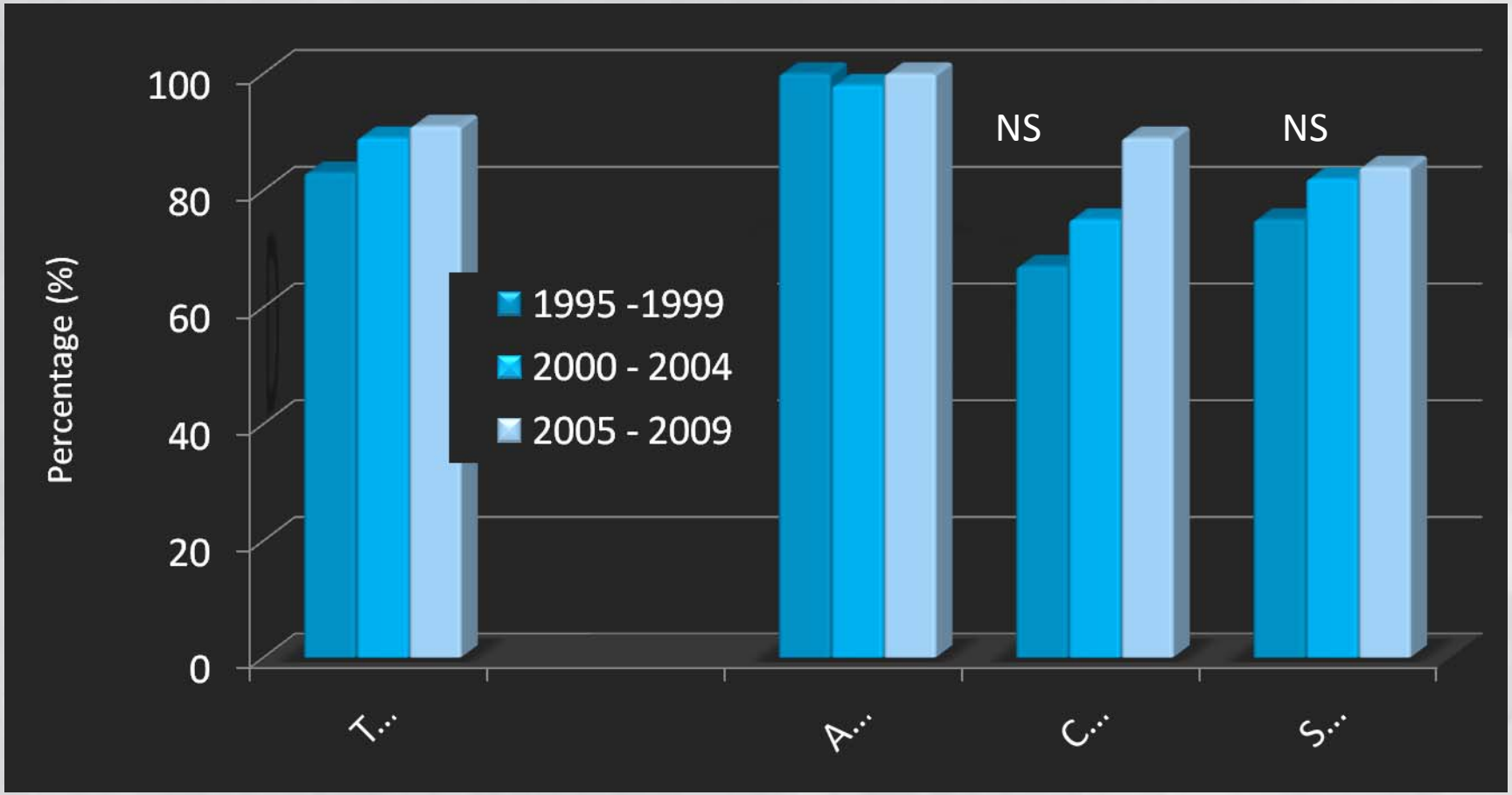
Evolution between 1995 and 2009.



D. Timbolschi, R. Favre, B. Doray. Fetal Diagn. Ther. 2014

Epidemiological datas

Percentage of prenatal diagnosis between 1995 and 2009.



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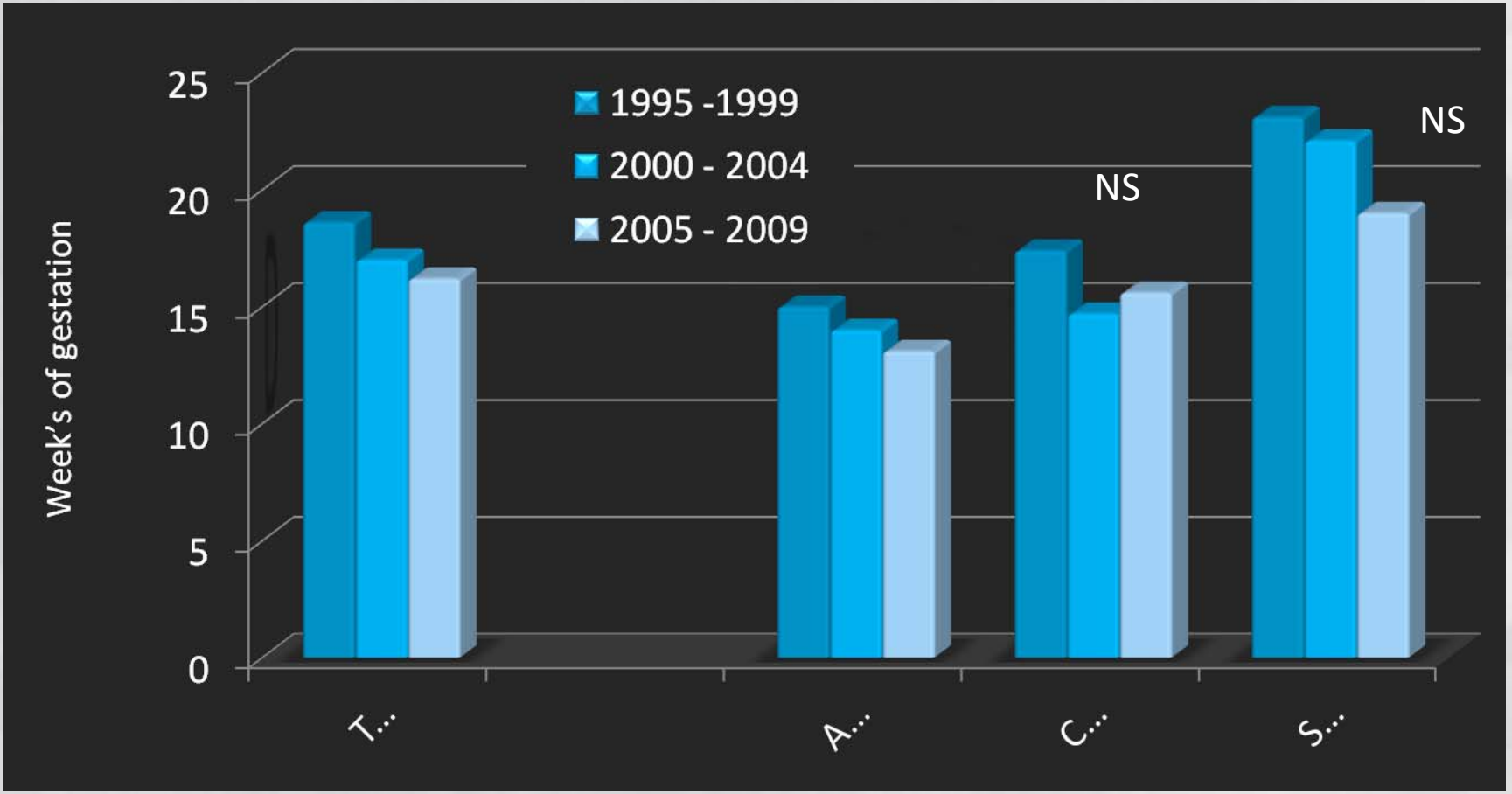
Epidemiological datas

Gestational age of prenatal diagnosis between 1995 and 2009.



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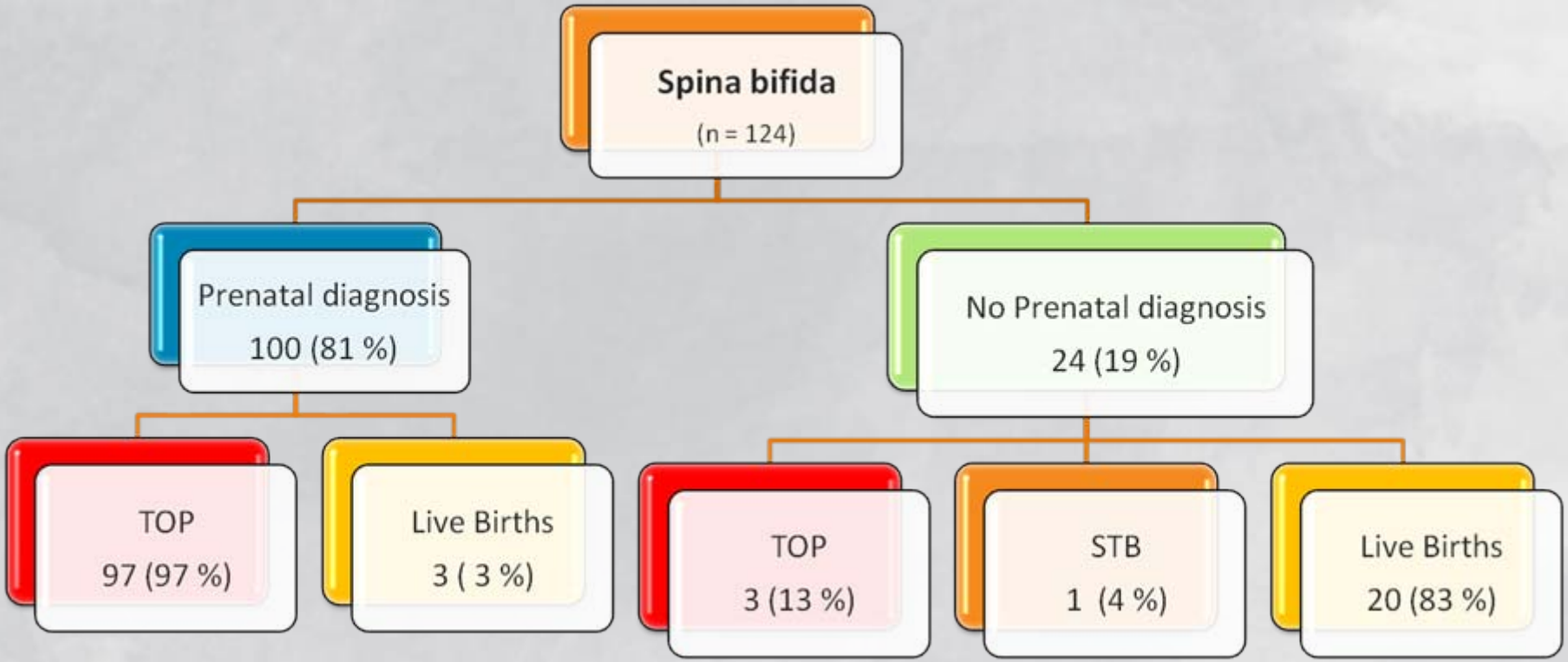
Epidemiological data

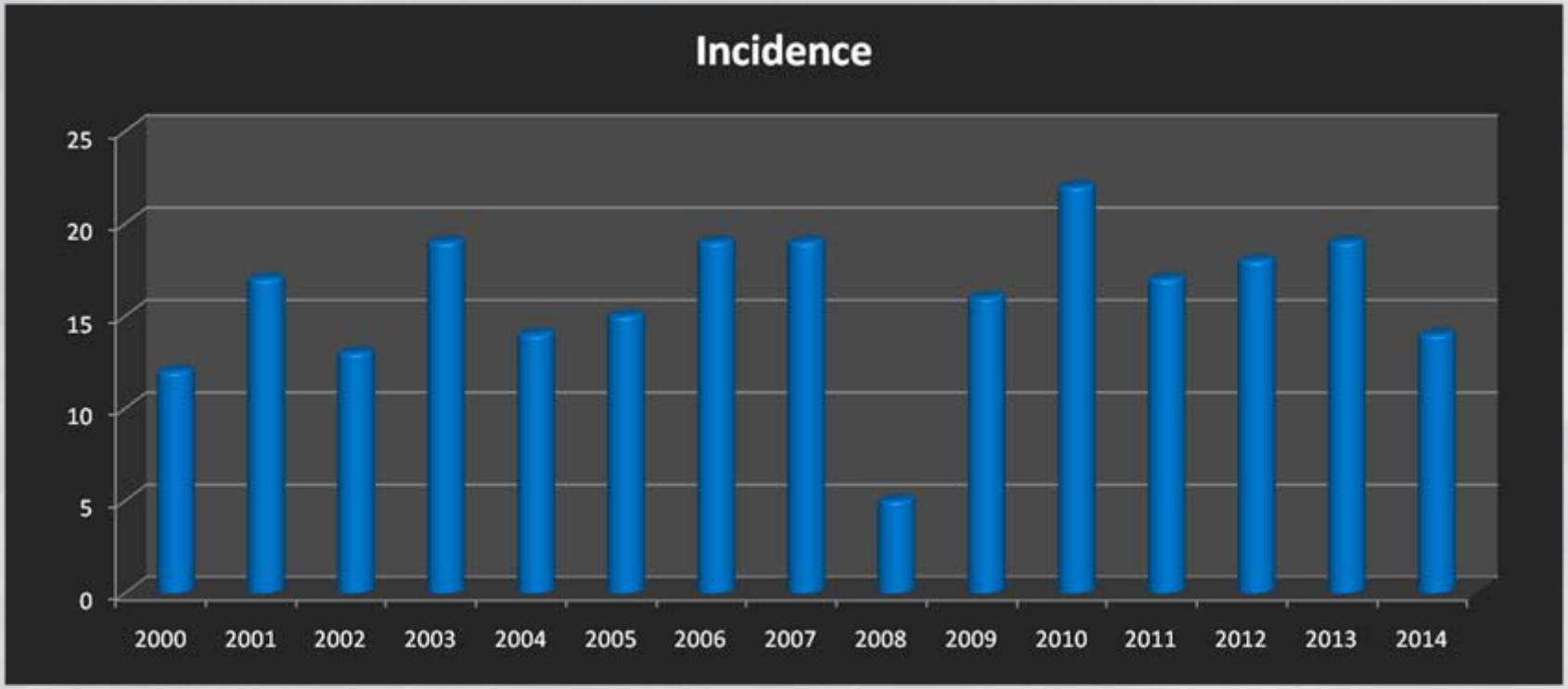
Outcome of spina bifida.



D. Timbolschi, R. Favre, B. Doray. Fetal Diagn. Ther. 2014

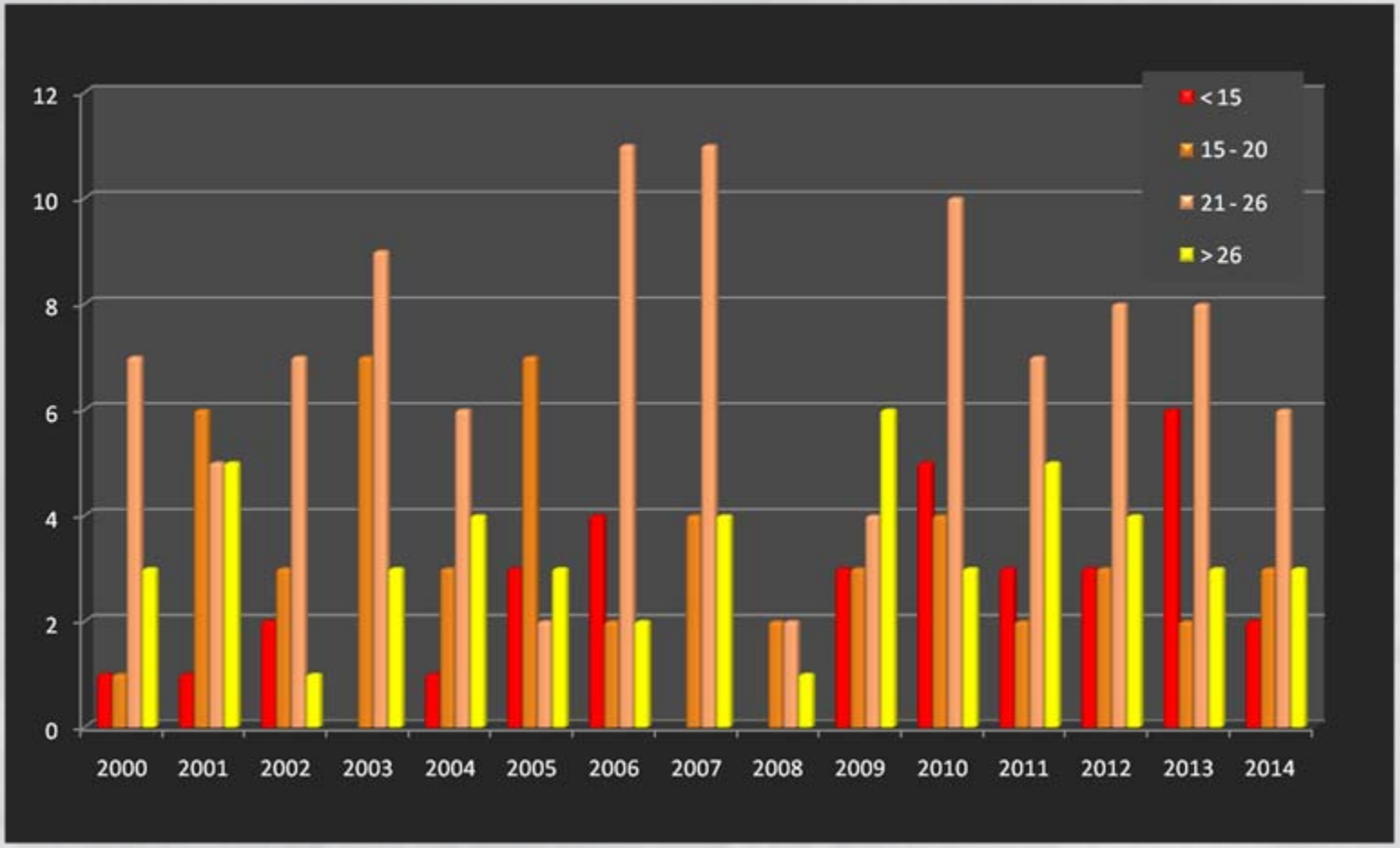
Data from CMCO

- Retrospective study from data of the Prenatal Registry of the Fetal Medicine dept. between 2000 and 2014.
- Total of 69.754 Fetal Ultrasound
- 239 cases of NTD



Data from CMCO

- **Evolution of the gestational age at diagnosis**



Data from CMCO

- Maternal age : 29.9 ± 5.7 (15 – 43)
- Gestational age : 22.4 ± 6.1 (12 – 38)
  - < 15 week's 34 (14.2 %)
  - 16 – 20 52 (21.8 %)
  - 21 – 26 103 (43 %)
  - > 26 50 (20.9 %)
- Singelton : 228 (95 %)
- Twin : 11 (4.6 %)
- Fetal Caryotype
  - Normal 154 (90,5 %)
  - Trisomy 18 11 (6.5 %)
  - Triploidy 3 (1.8 %)
  - Structure abn. 2 (1.2 %)
- Ultrasound aspect :
  - Ventriculomegaly 90 (38 %)
  - Club foot 38 (16 %)
  - Syndrom 28 (11 %)

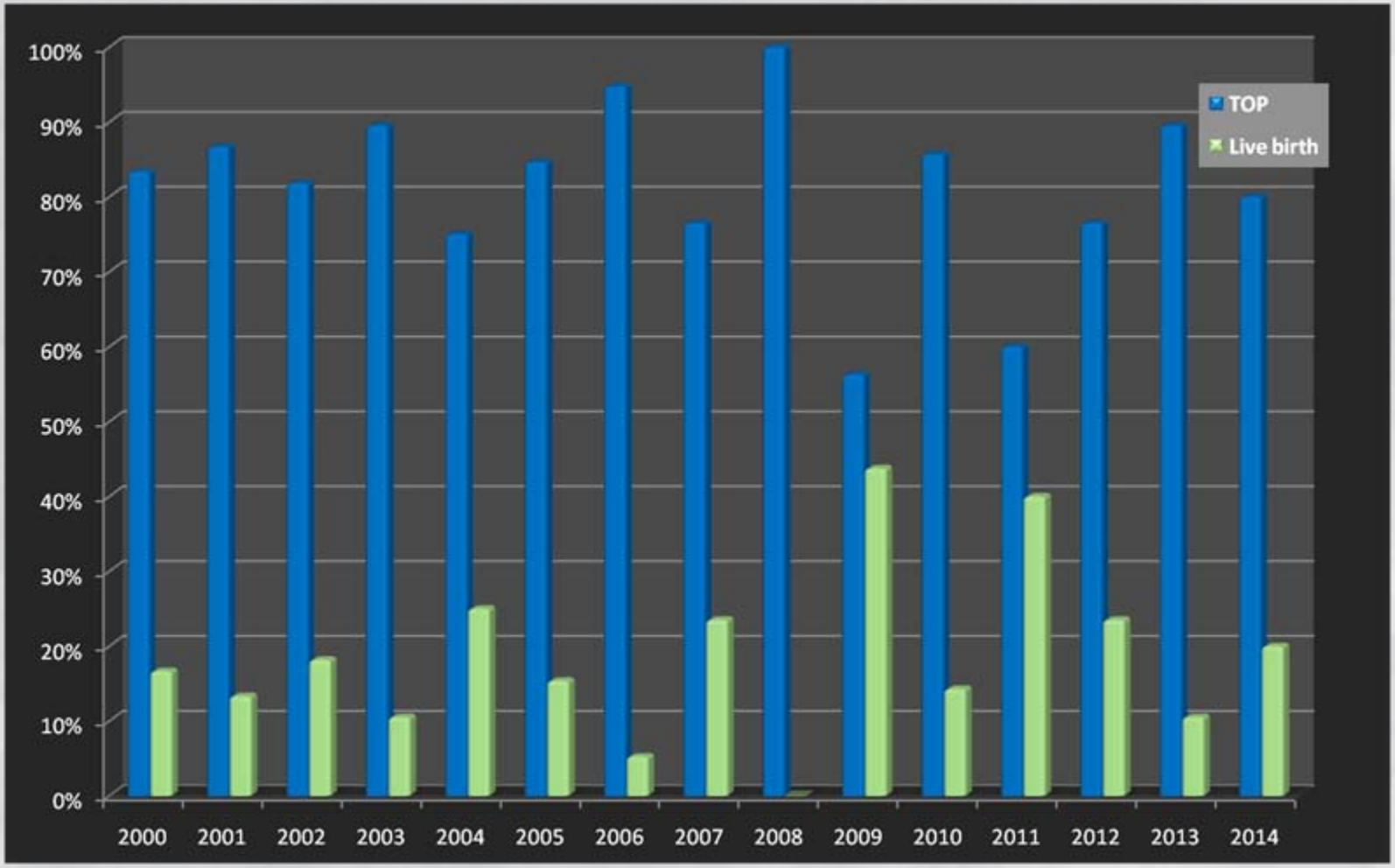
Outcome:

- TOP 178
- STB 6
- Live births 42 (17 %)
- Loss 13

- Neonatal death 13

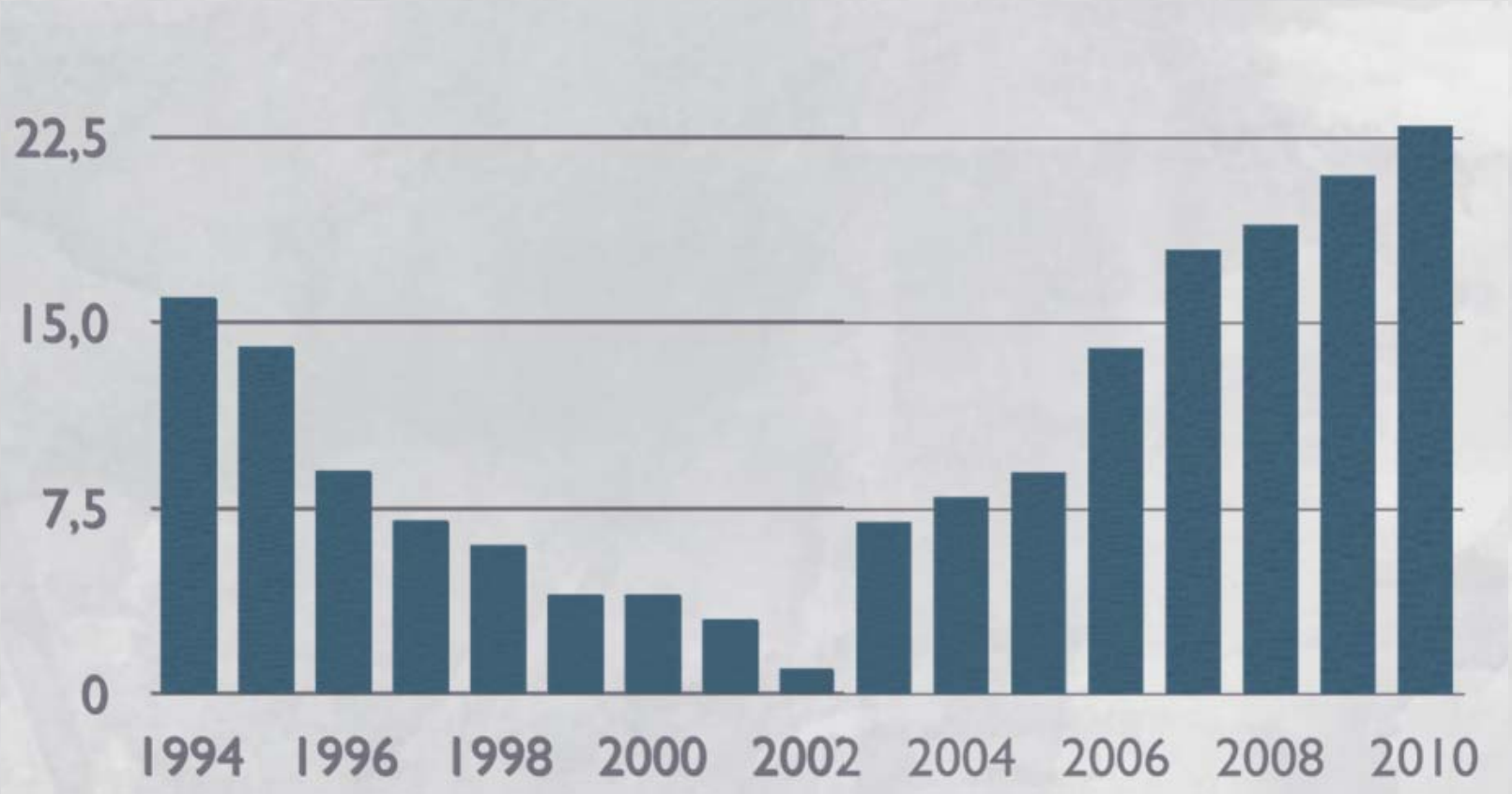
Data from CMCO

- **Evolution of the outcome**



Epidemiological datas

Evolution of the Neurosurgical activity between 1994 and 2010.



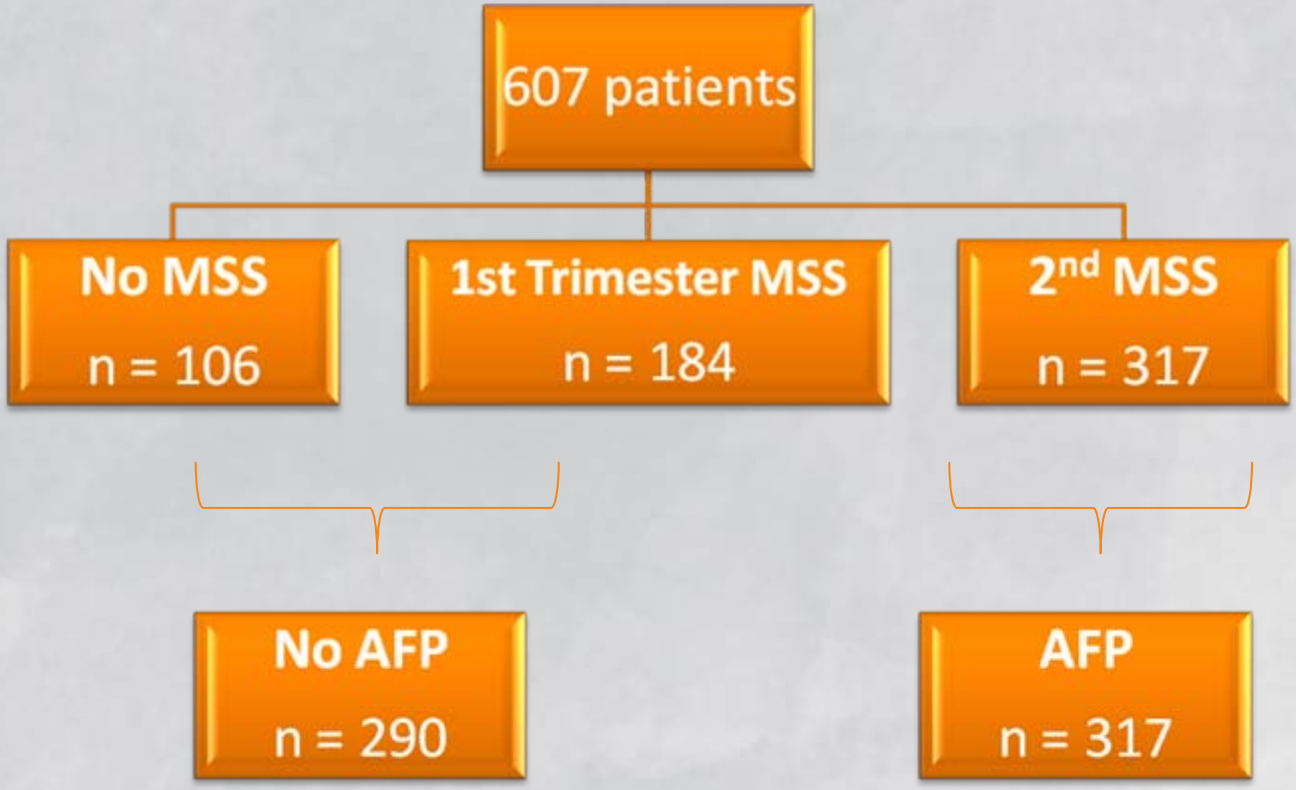
Michel Zerah. Dept of Pediatric Neurosurgery 2014

Impact of the loss of AFP

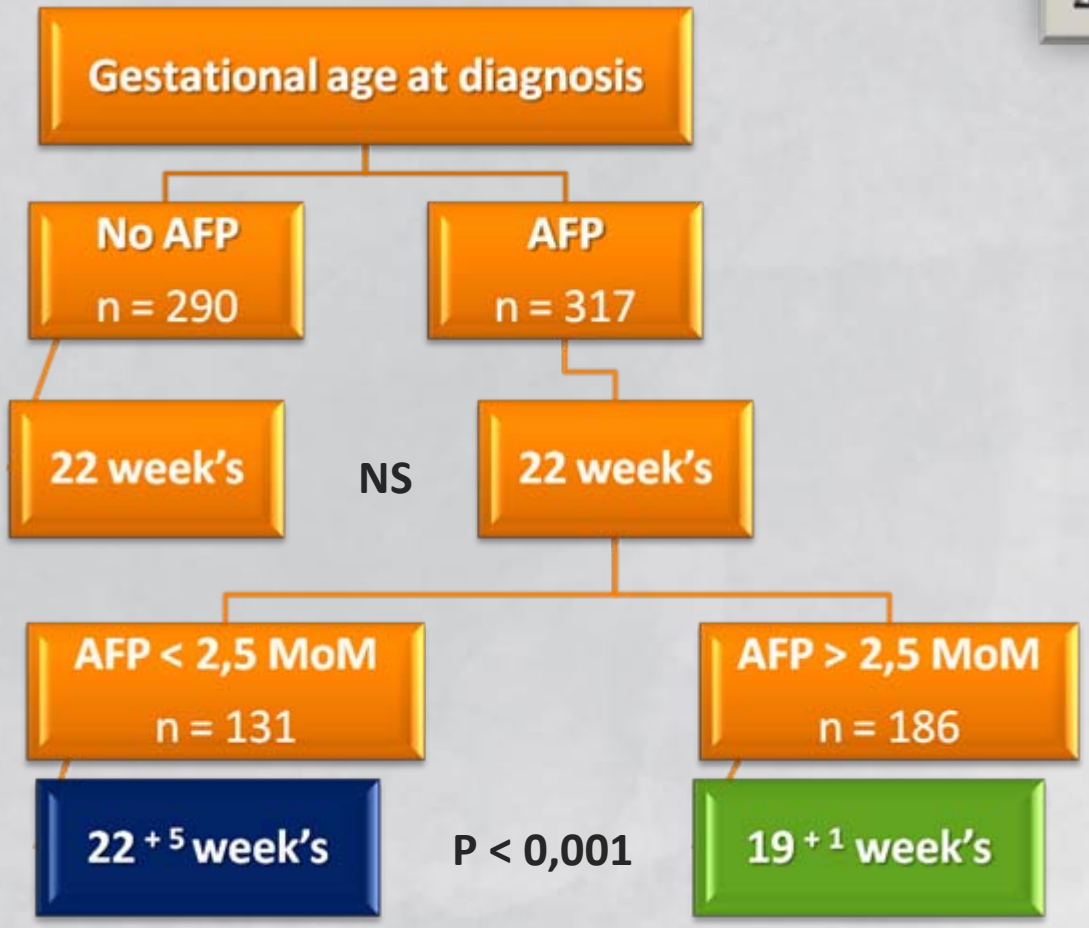
Objective: Evolution of the rate of prenatal diagnosis of spina bifida after the national modification of the use of the 2nd trimester MSS toward the 1st trimester.

- **Retrospective study from 2009 and 2011.**
  - **Comparison:**
    - **2009: AFP with the 2nd trimester MSS**
    - **2010 : Transitional period**
    - **2011 : No AFP, because of the use of 1st trimester MSS**
- **858 spina bifida – 190 loss to follow up**
- **Maternal age : 29 [15 – 52]**
  - **Previous NTD : 36**
  - **Maternal Epilepsy : 13**
  - **Diabetes : 9**
  - **Obesity (BMI > 35) : 58**
- **Aneuploidy : 25 (2.9 %)**
- **Syndrom : 141 (16.4 %)**
- **Isolated Spina bifida : 607**

Impact of the loss of AFP



Impact of the loss of AFP



Impact of the loss of AFP

Gestational age at diagnosis

- **Group without AFP or normal AFP (n = 131 + 290).**
  - **14 – 20 $^{+6}$: n = 69 (16.3 %)**
  - **21 – 26 : n = 307 (73 %)**
  - **> 26 : n = 36 (8.5 %)**

- **Group with AFP and AFP > 2.5 MoM (n = 186).**
  - **14 – 20 $^{+6}$: n = 120 (64.5 %)**
  - **21 – 26 : n = 60 (32.2 %)**
  - **> 26 : n = 6 (3.2 %)**

Impact of the loss of AFP

Outcome

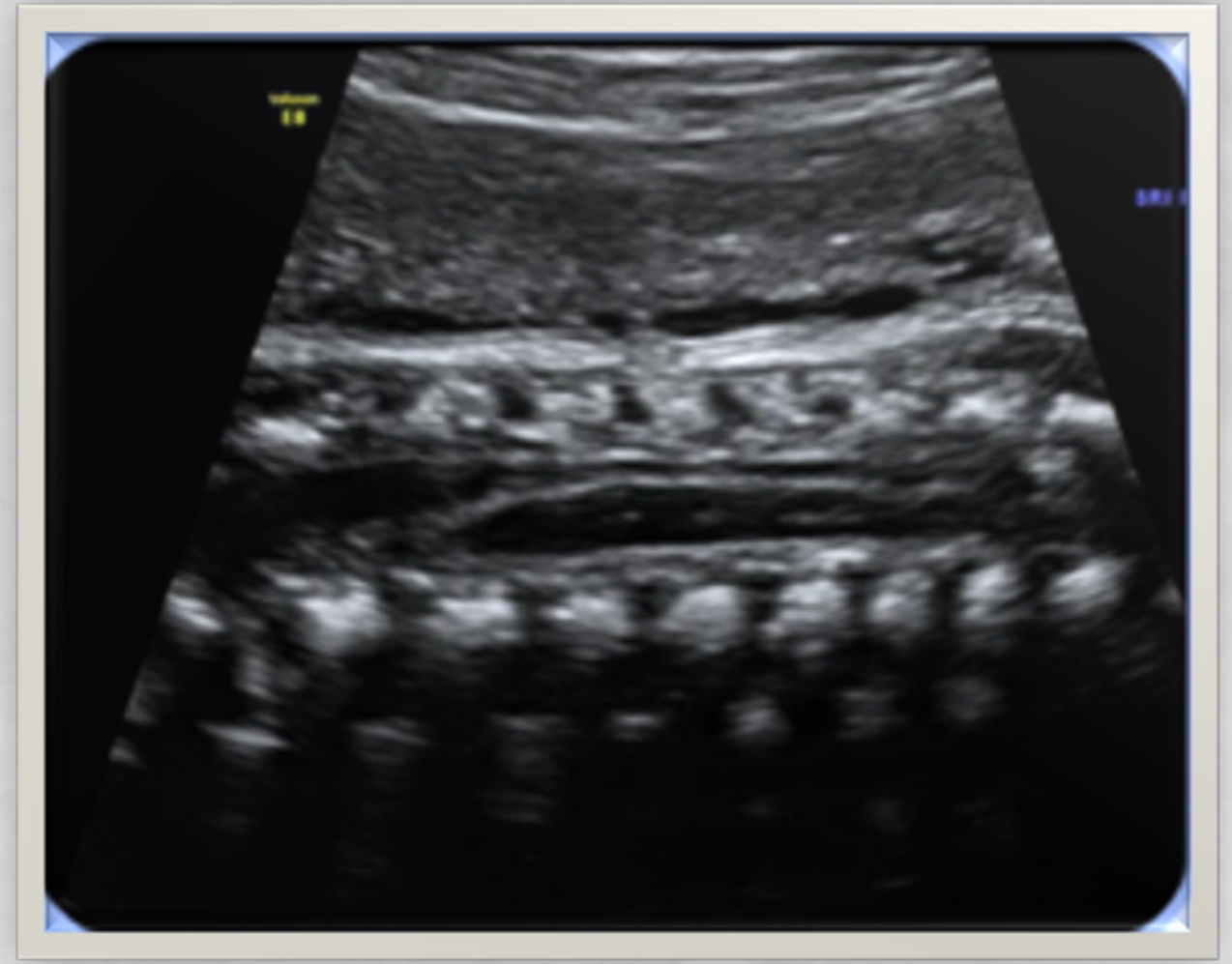
Live births : 70 (8.3 %) 1 FN at ultrasound

TOP : 768 (92.3 %)

Fetal loss : 4 (0.5 %)

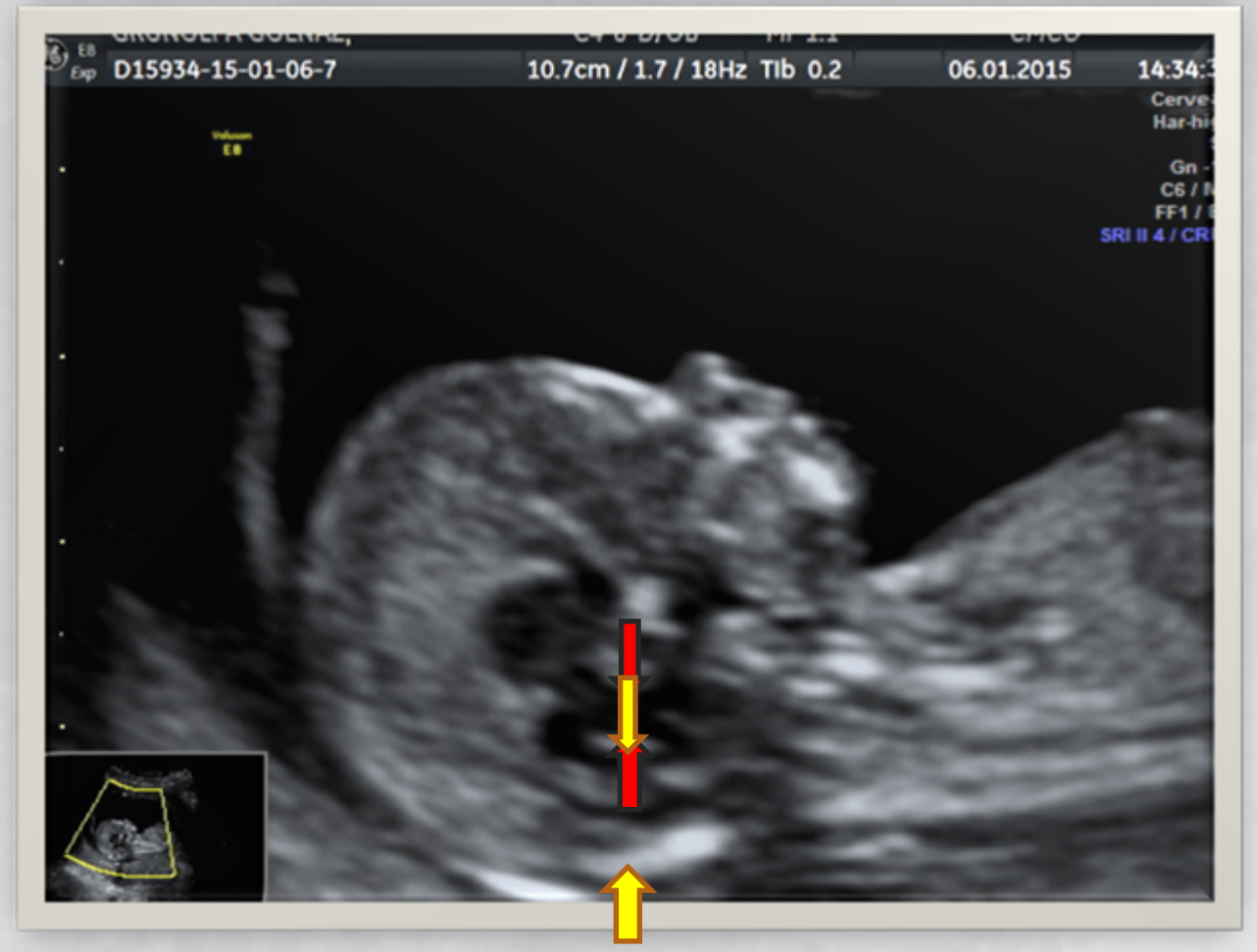
Syndrom : 166 98 % TOP

Prenatal screening



Prenatal diagnosis

Prenatal screening



Lachmann R & coll. Prenat Diagn. 2011

PRIUM Study


Hôpitaux Universitaires Paris Est
TROUSSEAU
LA ROCHE-GUYON
UPMC
PARIS
ASSISTANCE PUBLIQUE
HÔPITAUX DE PARIS
ETUDE PRIUM :
RÉPARATION PRÉNATALE DES MYÉLOMÉNINGOCÈLES
Investigateur coordonnateur : Pr Jean-Marie JOUANNIC
Responsable Scientifique : Pr Michel ZERAH
Chef de Projet URC Paris Necker-Cochin : Laurence Lecomte
Attachée de Recherche Clinique URC Paris Necker-Cochin : Elsa Bournaud
Statisticien : Pr Jean-Marc TRELUYER, Hôpital Necker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