

# **La diagnosi prenatale:**

## **finalità, utilizzo, comunicazione ai genitori**

**Patrizia Vergani**  
**Monza**



# Test di screening

- Ecografia
- Test prenatali per la identificazione della Sindrome di Down

# Ecografia

- Relativamente sicura
  - Non invasiva
- Facilmente disponibile



# Utilità dell'ecografia

- Esame diagnostico
  - ⇒ “su indicazione”
- Esame di screening
  - ⇒ “di routine”



# Test di screening e/o test diagnostico

- Cochrane 2001
  - Metanalisi di 8 studi clinici randomizzati (1982-1998) confronto routine vs indicazione
  - Solo 2 trial ne valutano l'efficacia rispetto alla identificazione dei feti malformati
    - » Helsinki trial -1886/87-
    - » RADIUS trial -1987/91-

# Helsinki trial

64 centri / 9310 partecipanti

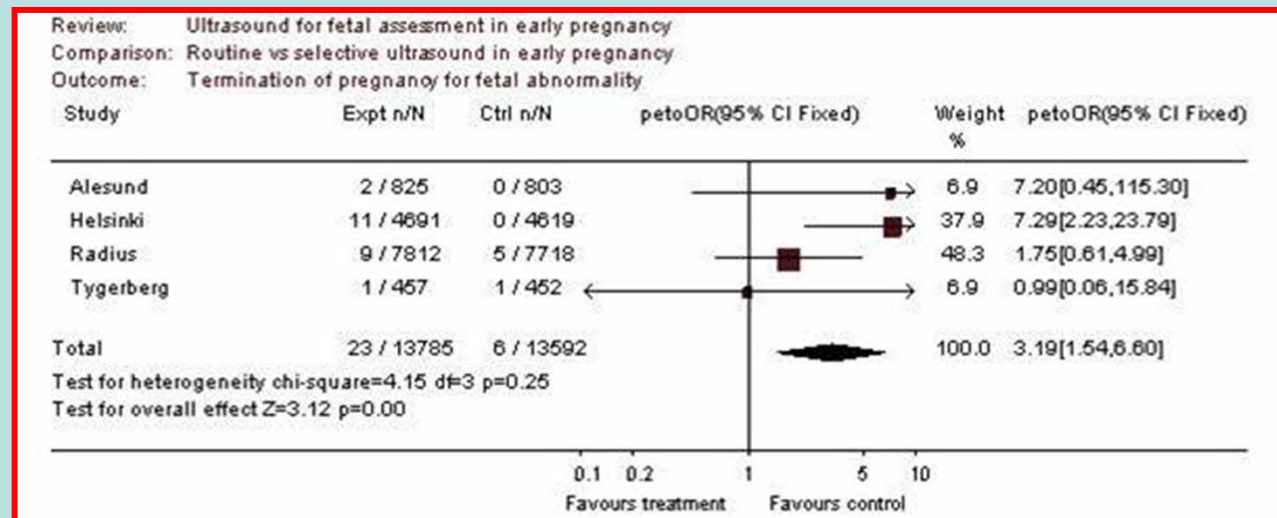
	Routine 16-20 w 4691	Su indicazione 4619
Pz sottoposte	100%	77%
N° scan	2,1	1,8
Malformazioni identificate	12/30 (40%)	13/48 (27%)
IVG	11	0
MP	4.5‰	8.4‰

# RADIUS trial

92 ostetrici / 15.530 partecipanti

	Routine 7812	Su indicazione 7718
Pz sottoposte	97%	45%
N° scan	2,2	0,6
Malformazioni identificate	65/187 (35%)	18/168 (11%)
Entro la 24 w	31 (17%)	9 (5%)
IVG	9	4
MP	6.3‰	5.1‰

# Ecografia di routine vs indicazione



30 anni di esperienza medica in una disciplina che propone continuamente

- La rinuncia terapeutica
- L'eutanasia degli individui “imperfetti”

## **Rinuncia terapeutica:**

Rottura Prematura delle Membrane Prima della  
Vitalità Fetale

- Rischi
  - Materni: Infezioni ➡ Sepsi
  - Fetali: Ipoplasia Polmonare ➡ Morte
- Terapia
  - Antibiotici?
  - Prevenzione e cura?

**NO: ABORTO “INEVITABILE”**

e conseguente induzione medica dell'aborto

## Terapia:

### Conduzione conservativa

#### Conservativa

antibiotici e cortisonici

114 casi: 2 IVG

112 casi ➡ 103 parto > 25 sett.

103 casi ➡ 49 vivi (47%)

**Vivi 49/114 (43%)**

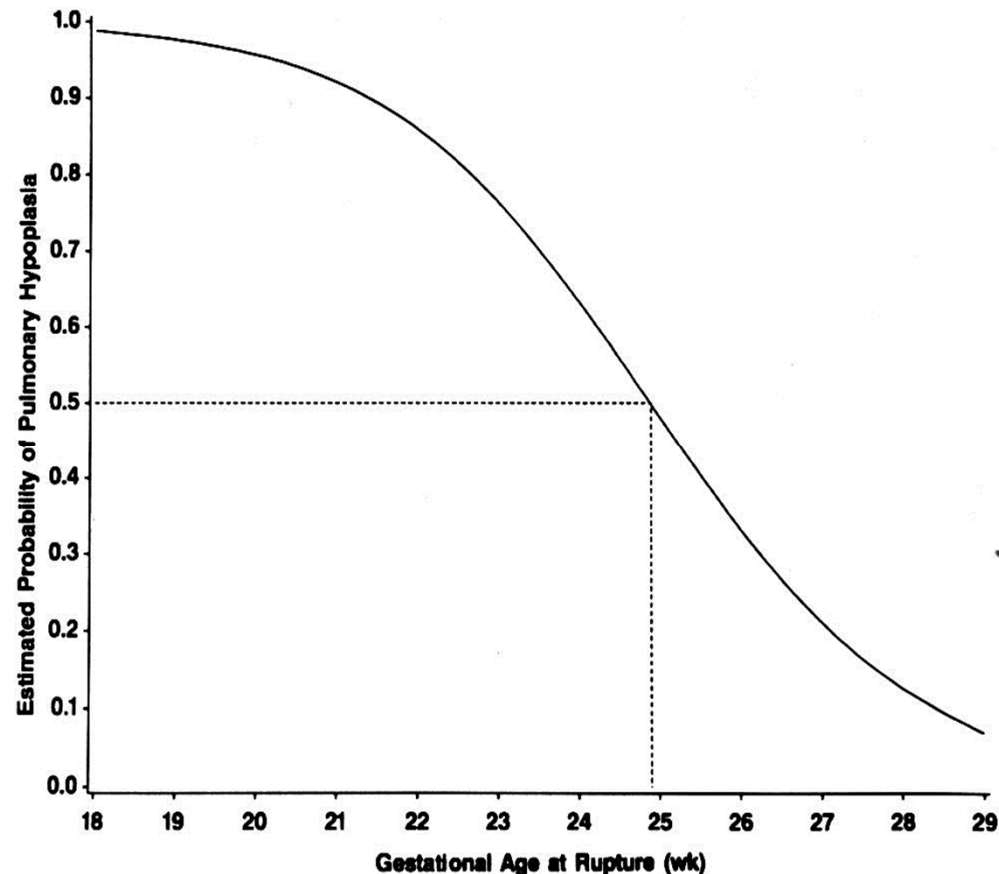
Sepsi 0%

<b>Rinuncia terapeutica:</b>	<b>vivi 0%</b>
<b>Terapia:</b>	<b>vivi 43%</b>

*Vergani P. et al*

## Risk Factors for Pulmonary Hypoplasia in Second-Trimester Premature Rupture of Membranes

*Am J Obstet Gynecol 1994;170:1359-1364*



- L'incidenza di ipoplasia polmonare è in relazione all'età gestazionale della rottura delle membrane

ma

solo nel sottogruppo con  
oligoidramnios  
in rapporto al periodo di  
latenza

- Delle gravidanze con una tasca di liquido amniotico >2 cm nessun neonato ha avuto ipoplasia polmonare

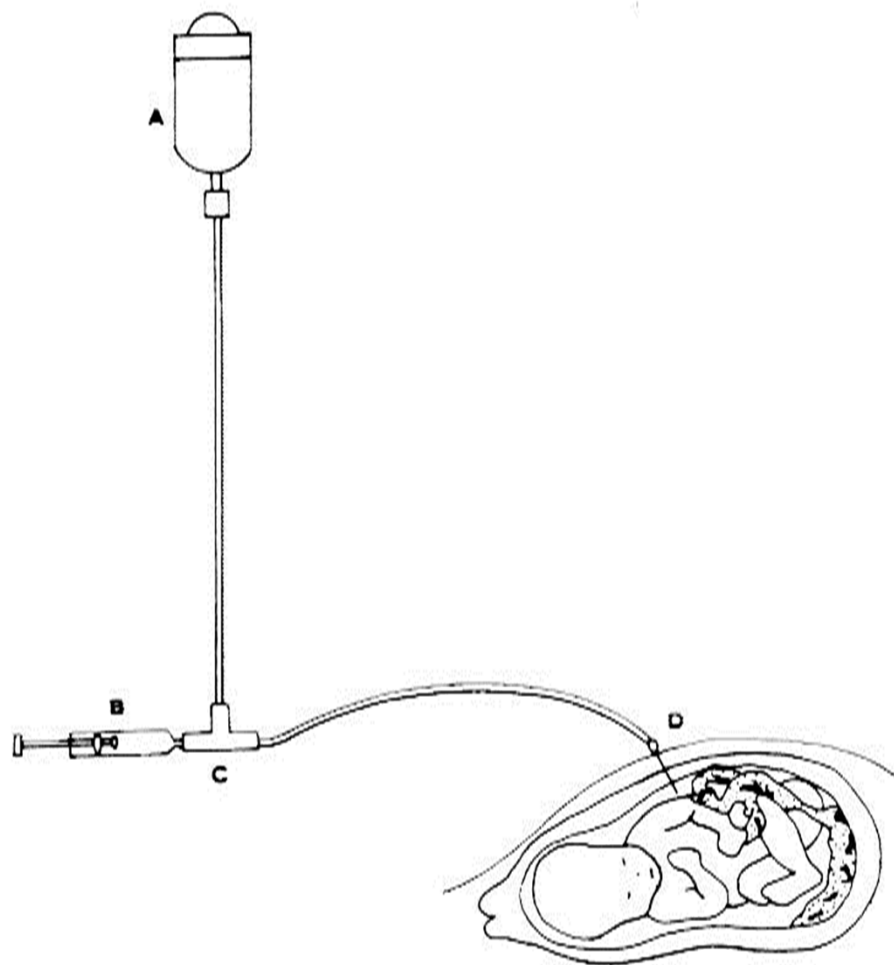


# Amnioinfusione

- Poichè il volume del liquido amniotico gioca un ruolo critico sull'esito della gravidanza, abbiamo intrappreso uno studio relativo alla possibilità di ripristinare il liquido amniotico

## Amnioinfusione transaddominale

***Vergani et al: Am J Obstet Gynecol 1996;175:465***



**Locatelli A; Vergani P; Di Pirro G; Doria V; Biffi A; Ghidini A.  
Am J Obstet Gynecol 2000;183:878**

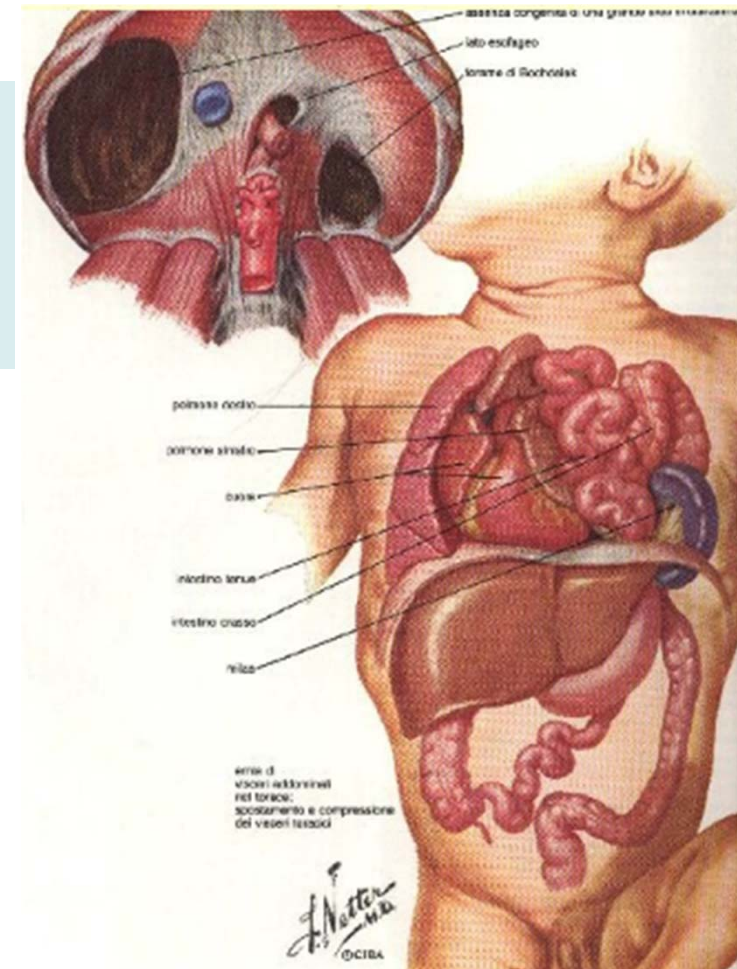
## **Role of amnioinfusion in the management of premature rupture of the membranes at <26 weeks' gestation**

	<i>Persistent oligohydramnios (n = 25)</i>	<i>Statistical significance</i>	<i>Amnioinfusion not necessary (n = 13)</i>	<i>Statistical significance</i>	<i>Successful amnioinfusion (n = 11)</i>
Gestational age at delivery (wk, median and range)	24.4 (17.0-29.0)	$P < .001$	28.5 (20.4-35.0)	$P = .8$	29.4 (22.0-35.3)
Birth weight <10th percentile (No.)	2 (8%)	$P = .5$	0	$P = .4$	1 (9%)
Pulmonary hypoplasia* (No.)	13/21 (62%)	$P < .001$	0/12	$P = .4$	1/10 (10%)
Postural deformities (No.)	4 (16%)	$P = .1$	0	$P = .2$	2 (18%)
Neonatal survival (No.)	5 (20%)	$P < .001$	12 (92%)	$P = .2$	8 (73%)
Neonatal follow-up (mo, median and range)	56 (12-96)	$P = .4$	53 (16-108)	$P = .6$	45 (12-106)
Abnormal neurologic outcome (No.)	3/5 (60%)	$P = .015$	0/12	$P = .4$	0/8

\*Includes patients in whom pathologic examination was diagnostic.

# Ernia diaframmatica

Evoluzione tecnica  
Terapia chirurgica





LHR  
Area Toracica/ Circonferenza  
Cefalica

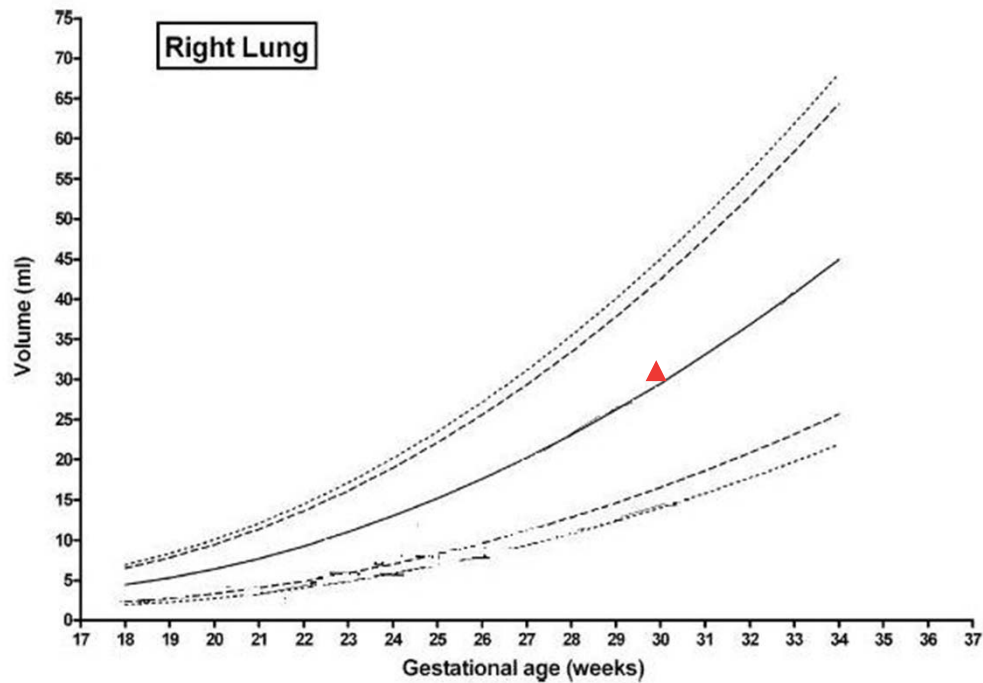
# LHR & CDH Outcomes

LHR	ECMO	SURVIVAL
<b>&lt; 1.0</b> (n=9)	<b>44%</b>	<b>11%</b>
<b>1.0 - 1.4</b> (n=16)	<b>50%</b>	<b>38%</b>
<b>&gt; 1.4</b> (n=9)	<b>44%</b>	<b>67%</b>

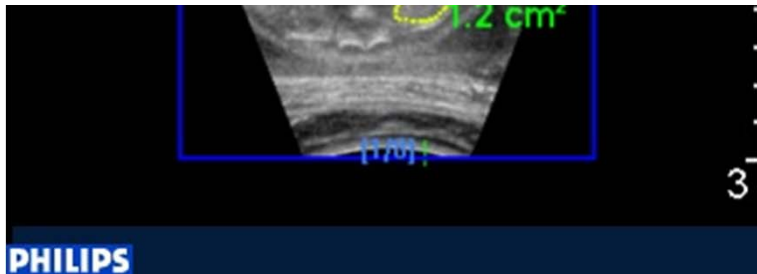
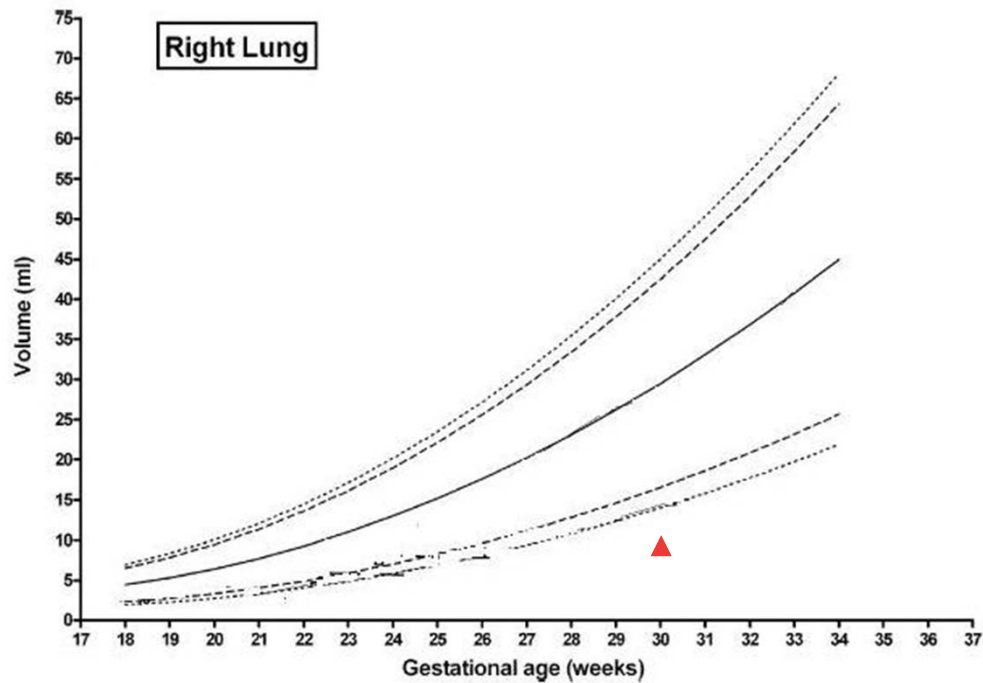
NB: LHR non funziona in assenza di erniazione epatica!!



# Volume polmonare a 30 SG

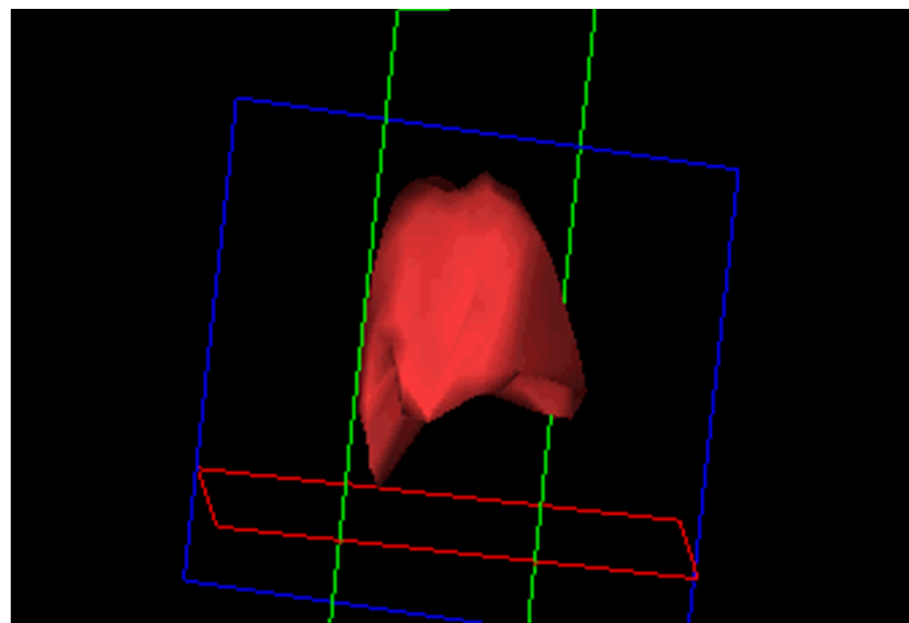
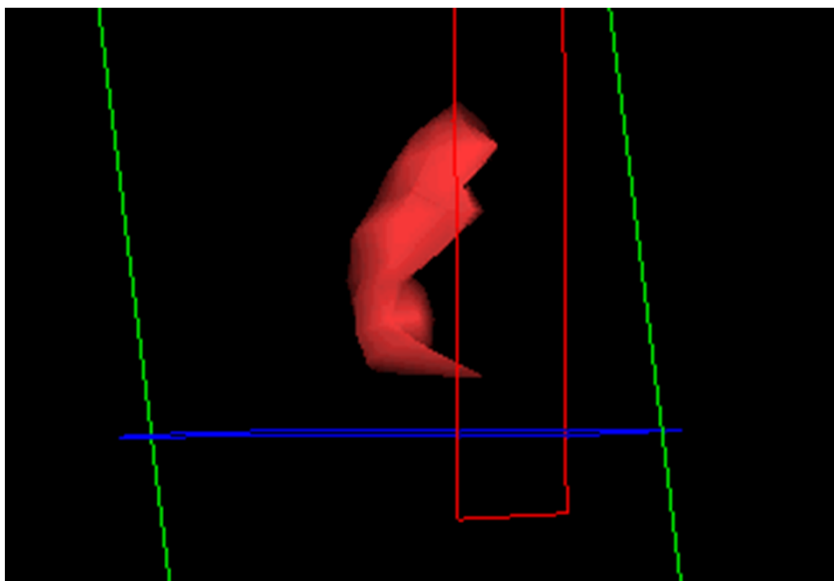


# Volume polmonare a 30 SG (CDH)

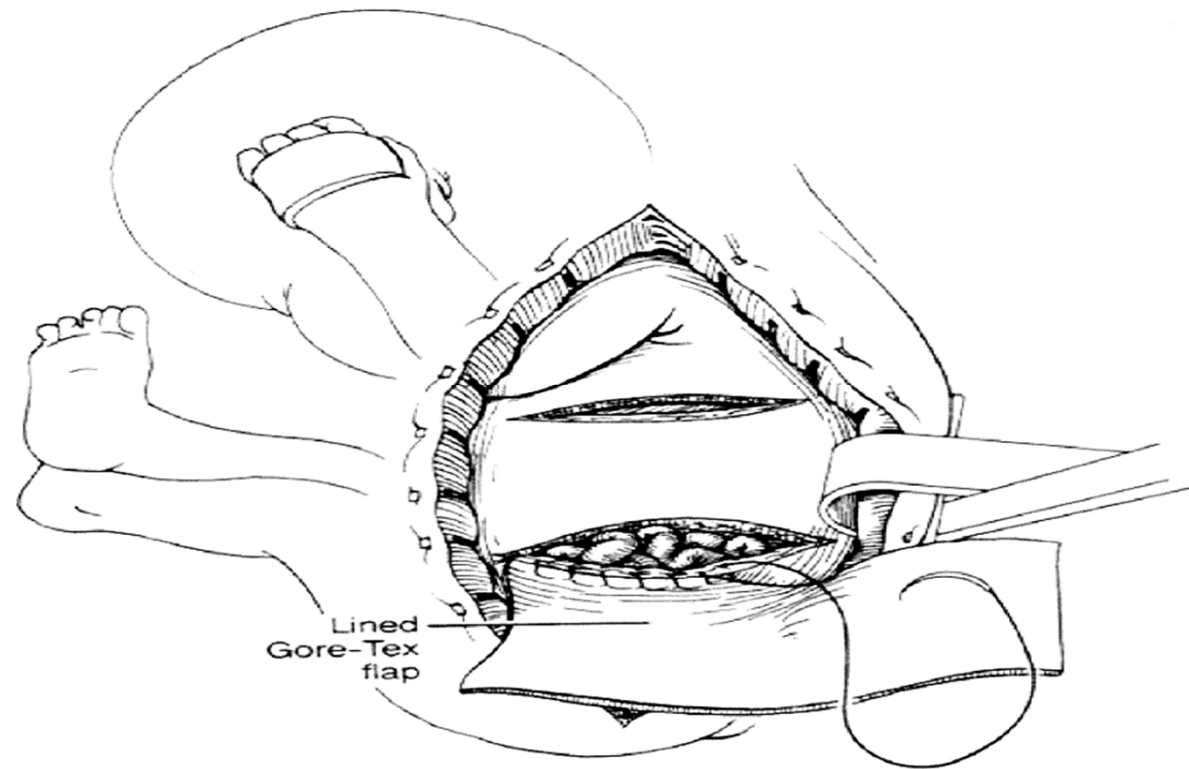


PHILIPS



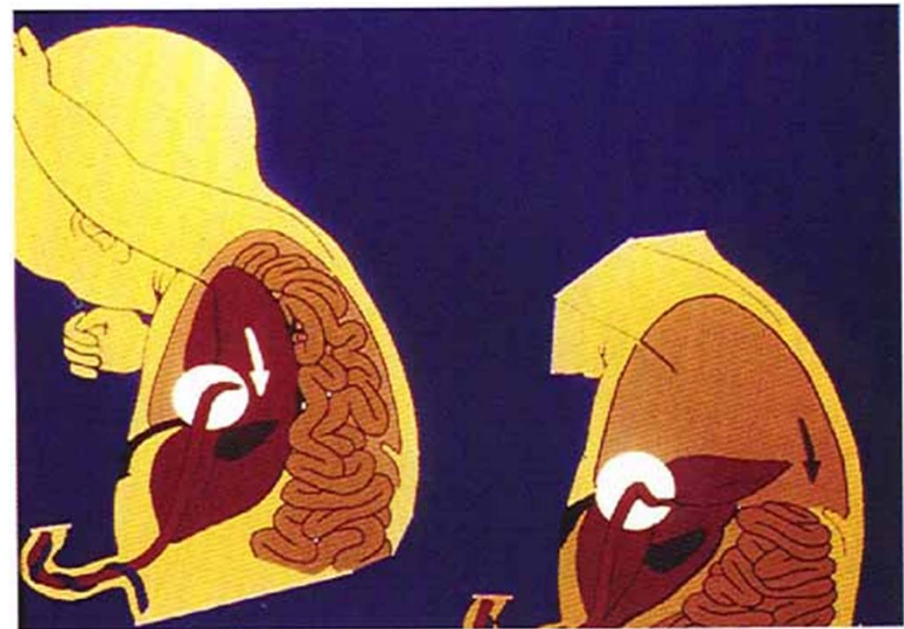


# ERNIA DIAFRAMMATICA CONGENITA (CDH)



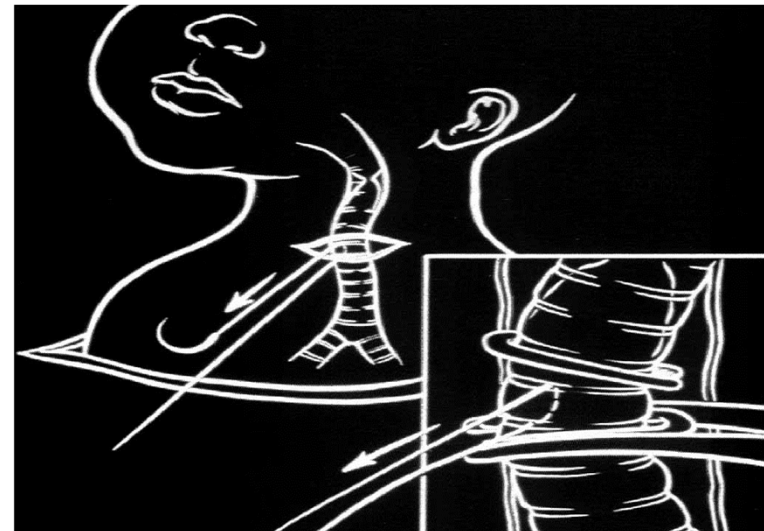
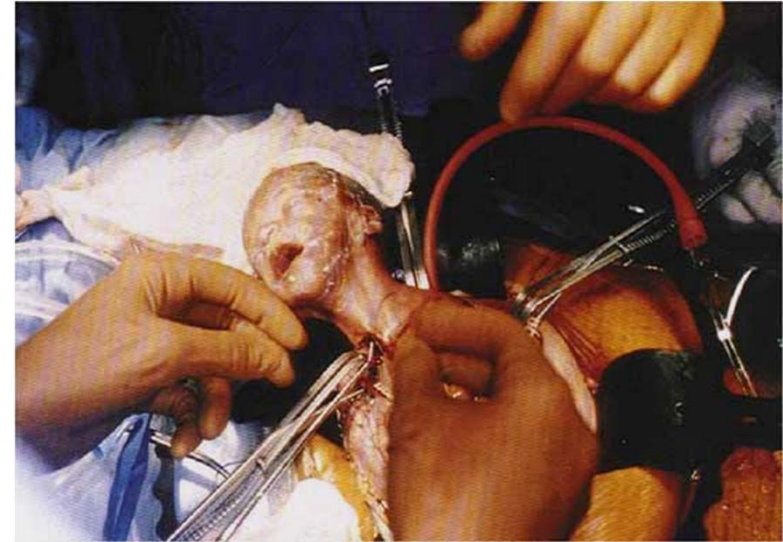
# CDH: Open fetal surgery

- Non migliora la prognosi nel sottogruppo di feti con fegato in addome, anche se eseguibile con buoni tassi di successo chirurgico (Harrison, J Pediatr Surg, 1997)
- 100% di mortalità nei casi con erniazione toracica del fegato, per ostruzione del flusso nella vena ombelicale al riposizionamento del fegato in addome (Harrison, J Pediatr Surg, 1993)



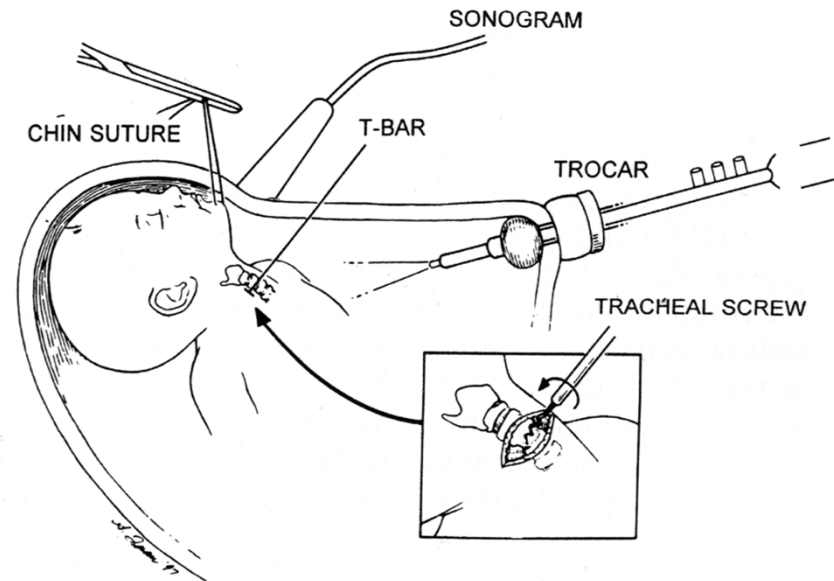
# CDH: Open tracheal occlusion (Harrison)

- Harrison(1997): 13 casi trattati
- 2casi: foam plug
- 11casi : clips
- Eg chirurgia:  $27 \pm 0.5w$
- Eg parto:  $30 \pm 0.6w$  (1 caso all'intervento)
- ECMO 2/13
- Sopravvivenza: 2/13 (15%)  
quasi tutte morti neonatali per complicanze legate a prematurità



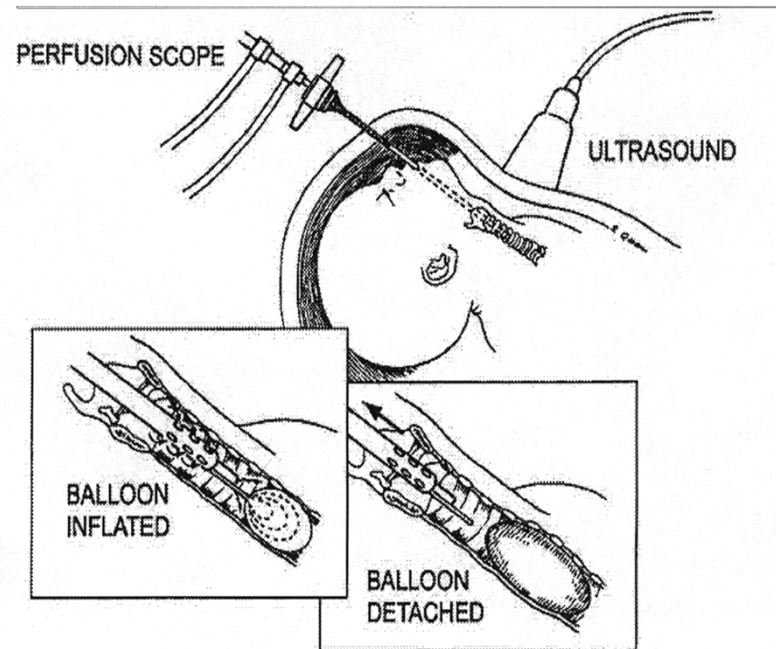
# CDH: Fetendo clip (Harrison)

- TECNICA: LPT, inserimento di un trocar da 10 mm e due da 5 mm (bloon-tip) sotto guida ecografica, irrigazione continua con salina, sutura sul mento del feto
- CASI: 8, EG diagnosi  $19 \pm 0.7w$ , LHR= $1.0 \pm 0.1$ , EG chirurgia  $29 \pm 0.5w$
- ESITI (Harrison): EG parto  $32 \pm 0.7w$ , ECMO 2/8 (25%), sopravvivenza 6/8 (75%), paresi corde vocali 2/6.



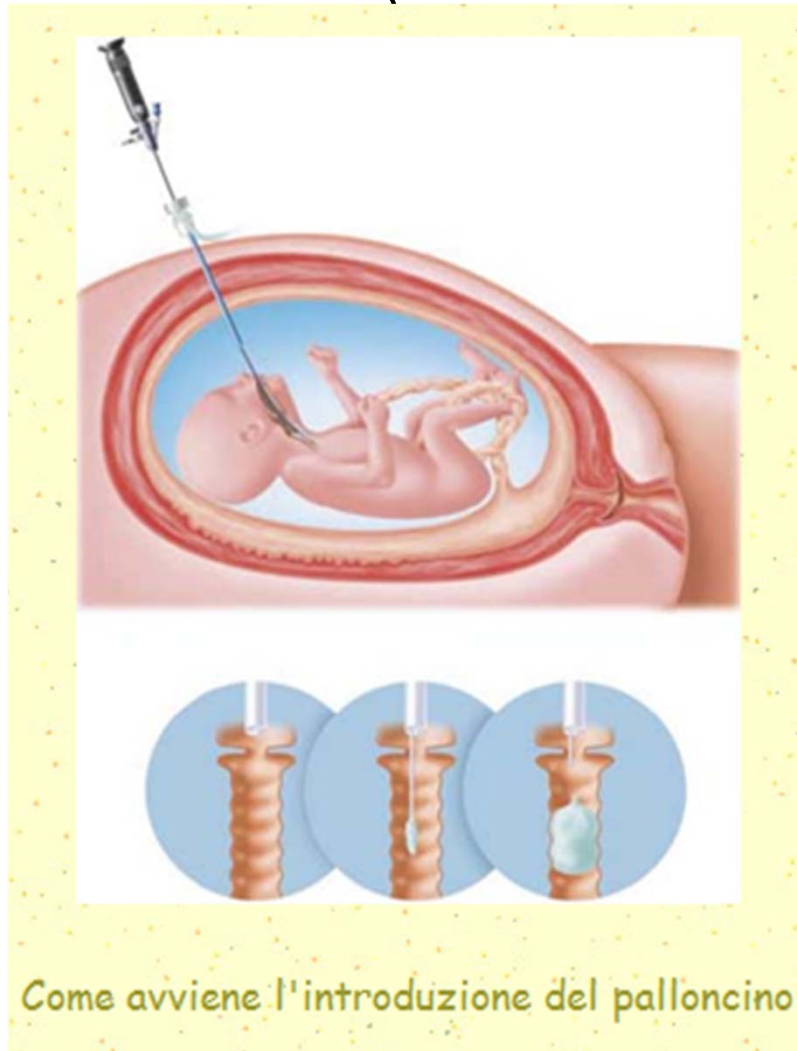
# CDH: Fetendo balloon occlusion

- Harrison, AJOG, 2001;185:730
- Due casi a prognosi sfavorevole
- Accesso con porta singola, con ecografica
- Sviluppo polmonare in utero 2/2
- Sopravvivenza 2/2



# TERAPIA PRENATALE DELL'ERNIA DIAFRAMMATICA SEVERA:

## F.E.T.O. (Fetal Endoscopic Tracheal Occlusion)



### **Indicato se LHR < 1**

Sopravvivenza passa da 10%  
al 60%

### **Posizionato**

Tra la 24 e 28 settimana e  
lasciato in sede almeno 6  
settimane

### **Rimosso**

tra la 32 e la 34 settimana

# Spina bifida

Dall'eutanasia alla nascita  
alla  
Terapia chirurgica ad utero aperto  
alla  
prevenzione primaria



# **SPINA BIFIDA**

**Paraplegia**

**Idrocefalo**

**Incontinenza**

**Disfunzioni sessuali**

**Deformità scheletriche**

**Ritardo mentale**

**'70 Lorber (UK) propone  
eutanasia passiva dei  
neonati**

**'80 screening (UK) con alfa-  
fetoproteina materna:  
aborto selettivo di tutti i  
feti affetti**



# SPINA BIFIDA

## Eutanasia passiva vs trattamento aggressivo:

Se la maggior parte dei bambini non trattati moriva in pochi mesi (Lorber), si era costretti a trattare i sopravvissuti tardivamente

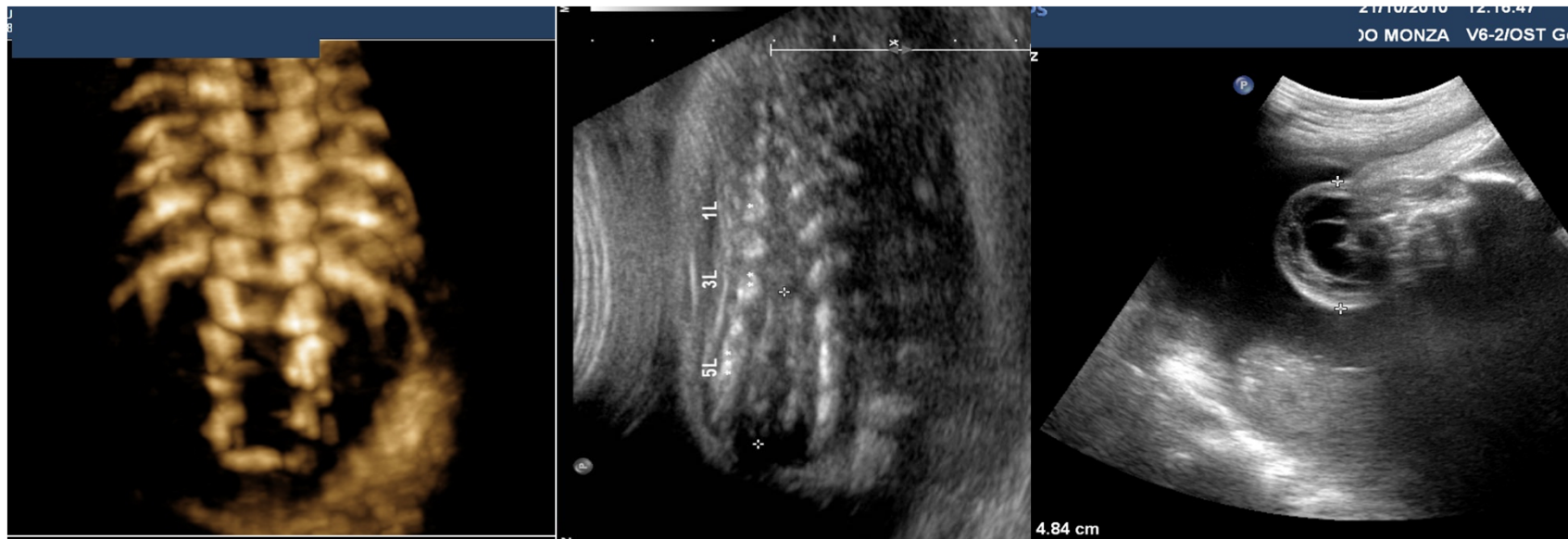
⇒ **aggravamento degli handicap neurologici ma soprattutto dell'idrocefalo con ritardo mentale**

Proposta di rivedere i protocolli di cura e trattamento aggressivo alla nascita di tutti i nati

⇒ **QI nei neonati con forme severe trattati 91 vs 104 (ns) delle forme lievi che non necessitano lo shunt**

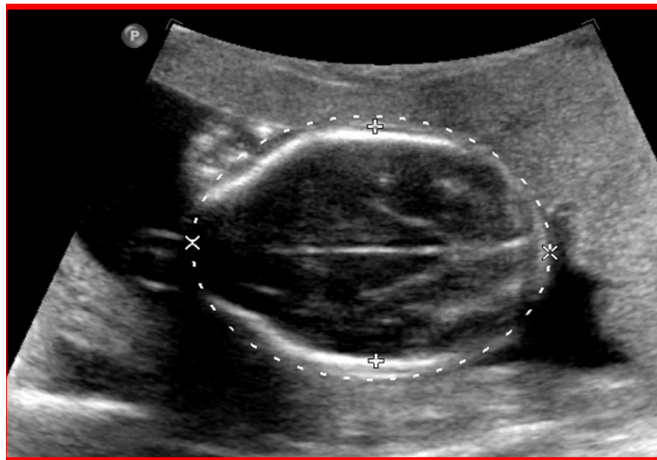
# Diagnosi prenatale di spina bifida

## Segni diretti



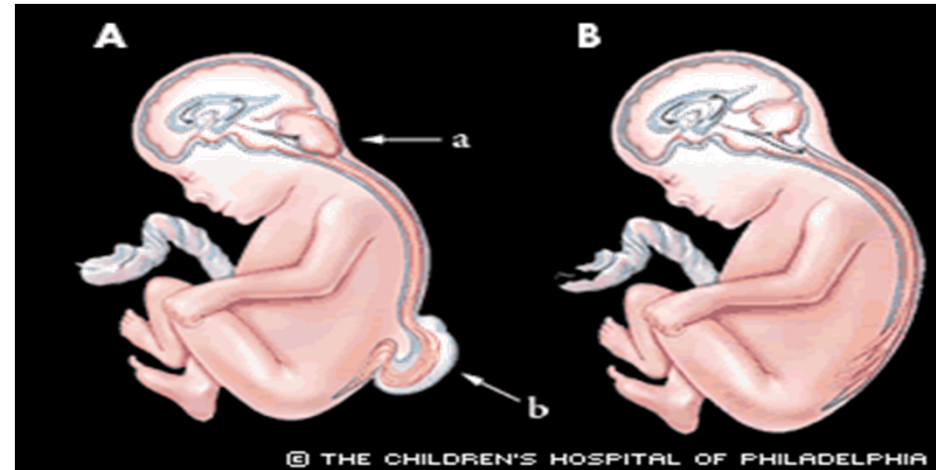
# Diagnosi prenatale di spina bifida

## Segni indiretti

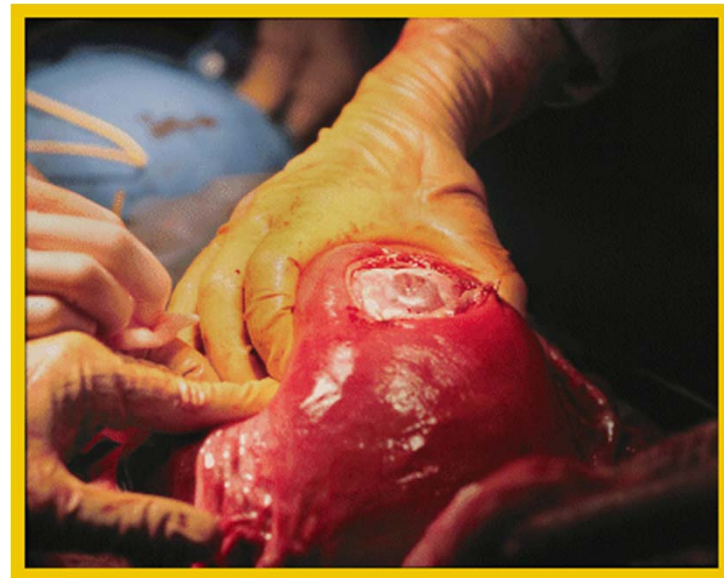


# APPROCCIO INVASIVO INTRAUTERINO

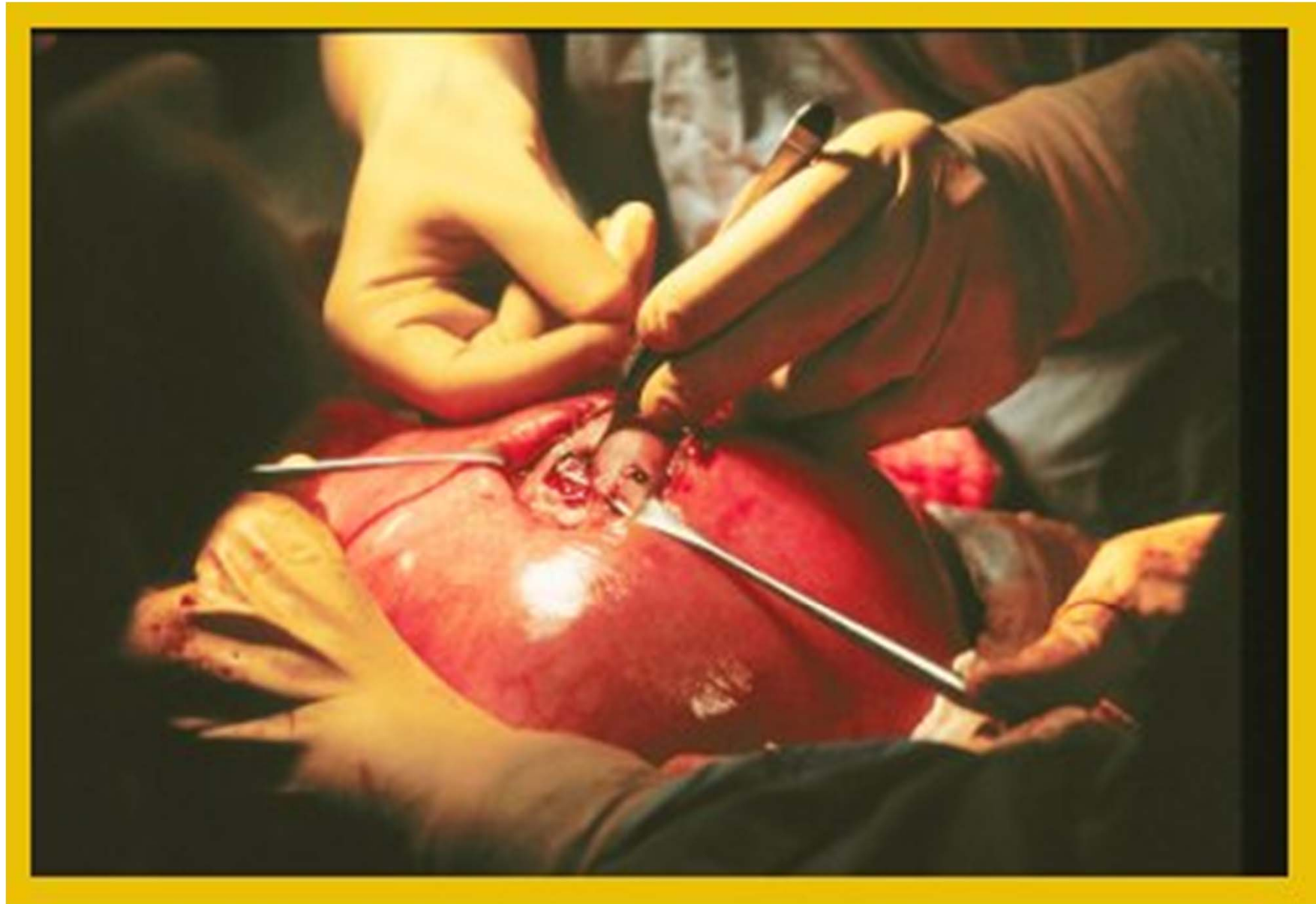
- Razionale:  
ridurre l'esposizione delle fibre nervose all'azione lesiva del liquido amniotico, operare a basse e.g.
- La correzione in utero riduce le alterazioni SNC secondarie (erniazione troncoencefalo, necessità di shunting)

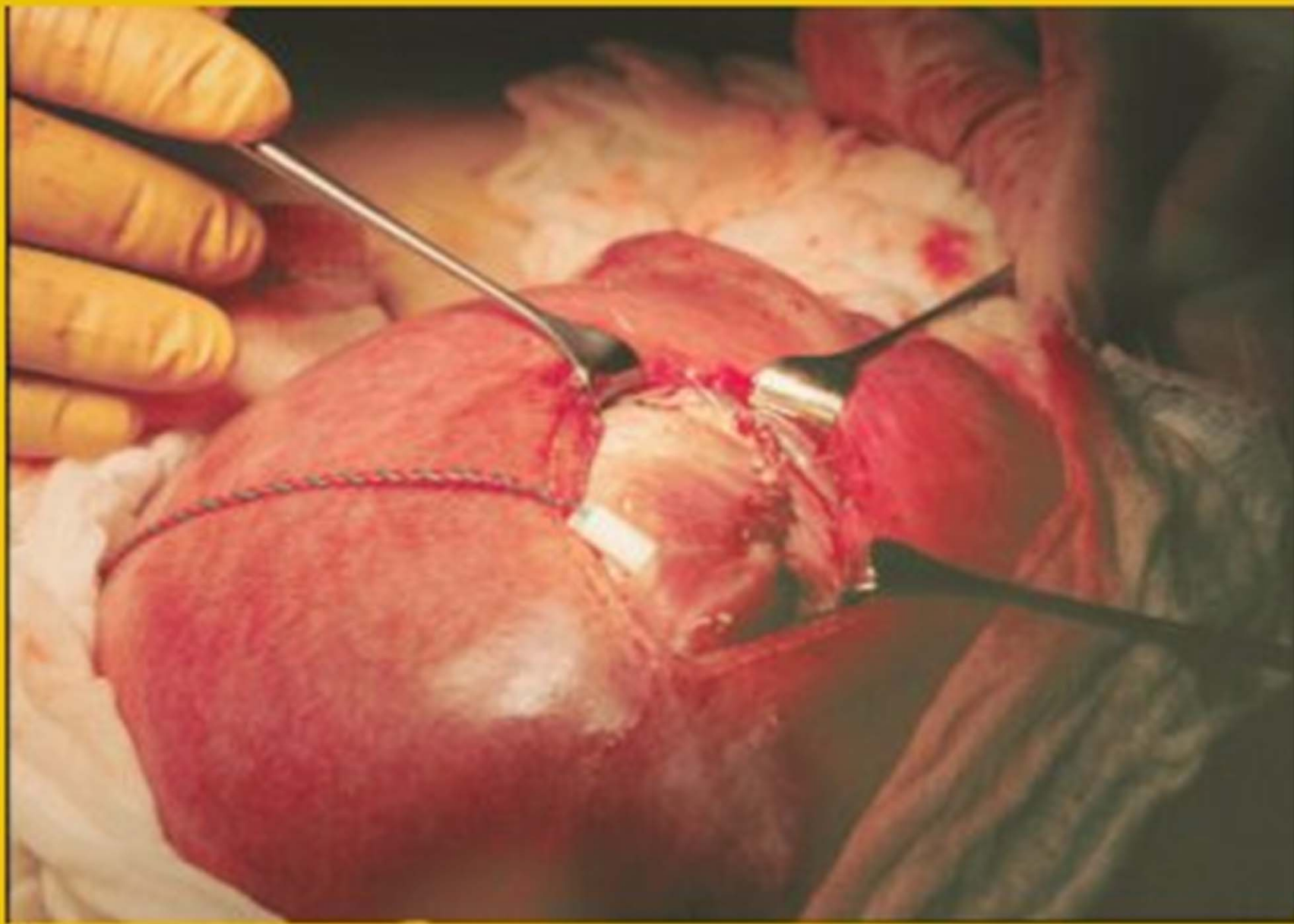


Hindbrain herniation before and after myelomeningocele surgery  
A. The hindbrain (a) protrudes into the spinal column because of the defect in the lower spine (b).  
B. Following early in utero surgical closure of the defect, the hindbrain moves back into a more normal position.













# Spina bifida ⇒ trattamento

**Adzick. Lancet 1998**

**Successful fetal surgery for spina bifida**

10 casi con grave erniazione del cervelletto

Operati tra la 22-25 settimana ⇒ 1 caso di morte

⇒ 1 solo caso di shunt VP

tutti con minori esiti neurologici vs gli attesi

**Bruner. JAMA 1999**

**Fetal surgery for myelomeningocele and the incidence of shunt-dependent hydrocephalus**

	<u>29 casi</u>	<u>23 controlli</u>
Shunt VP	59%	91%
Erniaz. Cervelletto	7%	45%
Piedi torti	28%	70%

# *The* NEW ENGLAND JOURNAL *of* MEDICINE

## A Randomized Trial of Prenatal versus Postnatal Repair of Myelomeningocele

N. Scott Adzick, M.D., Elizabeth A. Thom, Ph.D., Catherine Y. Spong, M.D., John W. Brock III, M.D.,  
Pamela K. Burrows, M.S., Mark P. Johnson, M.D., Lori J. Howell, R.N., M.S., Jody A. Farrell, R.N., M.S.N.,  
Mary E. Dabrowiak, R.N., M.S.N., Leslie N. Sutton, M.D., Nalin Gupta, M.D., Ph.D., Noel B. Tulipan, M.D.,  
Mary E. D'Alton, M.D., and Diana L. Farmer, M.D., for the MOMS Investigators\*

February 9, 2011, at NEJM.org.

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Society.*

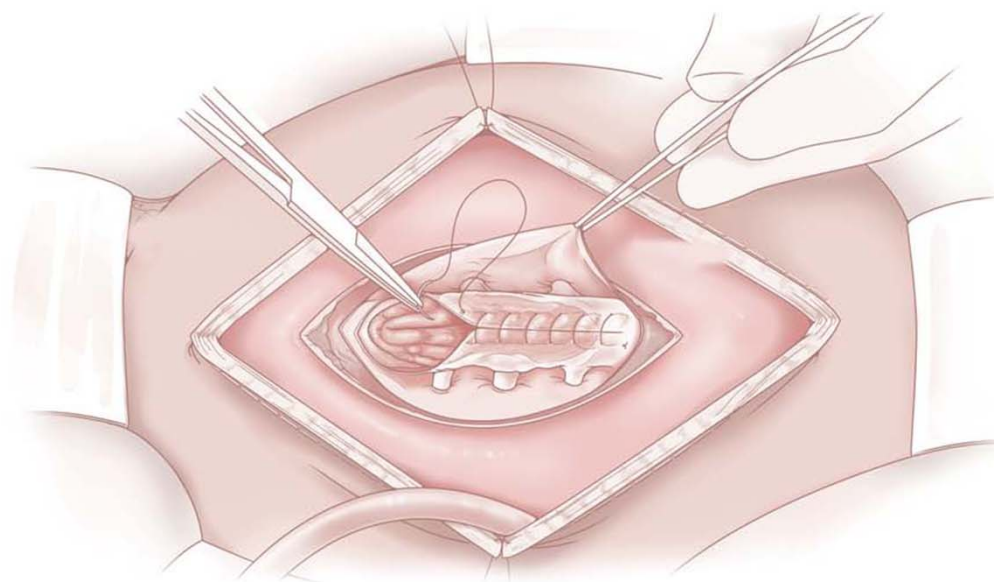
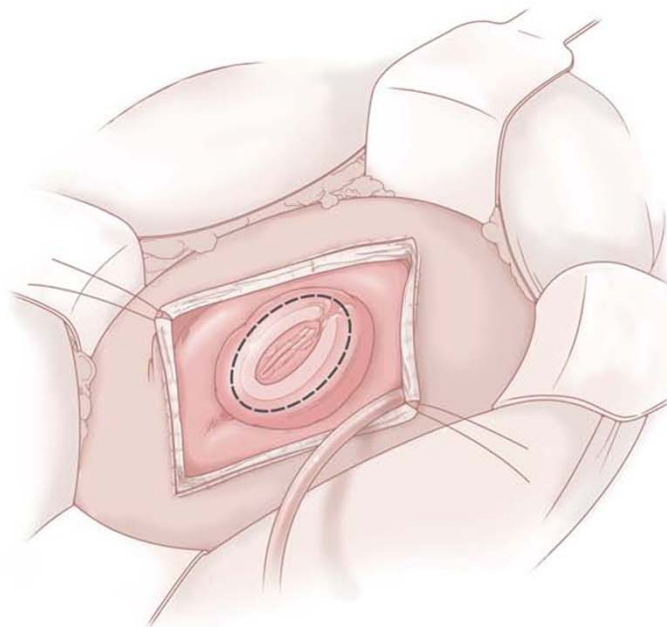
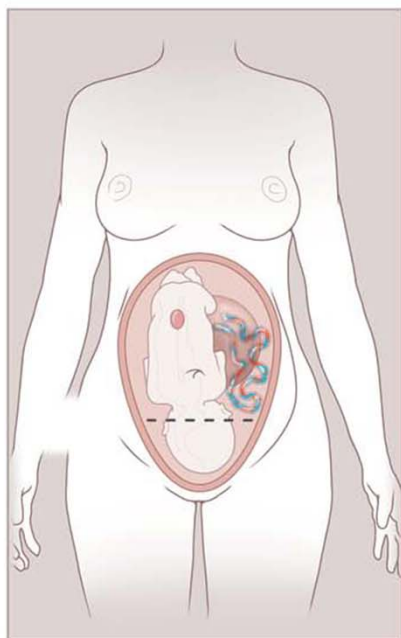


Table 3. Infant Outcomes at 12 Months.\*

Outcome	Prenatal Surgery (N=78)	Postnatal Surgery (N=80)	Relative Risk (95% CI)	P Value
Primary outcome — no. (%)	53 (68)	78 (98)	0.70 (0.58–0.84)†	<0.001
Components of the primary outcome — no. (%)				<0.001
Death before placement of shunt	2 (3)	0		
Shunt criteria met	51 (65)	74 (92)		
Shunt placed without meeting criteria	0	4 (5)		
Placement of shunt — no. (%)	31 (40)	66 (82)	0.48 (0.36–0.64)	<0.001
Any hindbrain herniation — no./total no. (%)	45/70 (64)	66/69 (96)	0.67 (0.56–0.81)	<0.001
Degree of hindbrain herniation — no./total no. (%)				<0.001†
None	25/70 (36)	3/69 (4)		
Mild	28/70 (40)	20/69 (29)		
Moderate	13/70 (19)	31/69 (45)		
Severe	4/70 (6)	15/69 (22)		
Any brainstem kinking — no./total no. (%)	14/70 (20)	33/69 (48)	0.42 (0.25–0.71)	<0.001
Degree of brainstem kinking — no./total no. (%)				0.001†
None	56/70 (80)	36/69 (52)		
Mild	4/70 (6)	8/69 (12)		
Moderate	7/70 (10)	17/69 (25)		
Severe	3/70 (4)	8/69 (12)		
Abnormal location of fourth ventricle — no./total no. (%)	32/70 (46)	49/68 (72)	0.63 (0.47–0.85)	0.002
Location of fourth ventricle — no./total no. (%)				<0.001†
Normal	38/70 (54)	19/68 (28)		
Low	28/70 (40)	29/68 (43)		
At foramen magnum	1/70 (1)	8/68 (12)		
Below foramen magnum	3/70 (4)	12/68 (18)		
Syringomyelia — no./total no. (%)	27/69 (39)	39/67 (58)	0.67 (0.47–0.96)	0.03
Epidermoid cyst — no./total no. (%)	2/67 (3)	1/66 (2)	1.97 (0.18–21.20)	1.00
Surgery for tethered cord — no./total no. (%)	6/77 (8)	1/80 (1)	6.15 (0.76–50.00)	0.06
Chiari decompression surgery — no./total no. (%)	1/77 (1)	4/80 (5)	0.26 (0.03–2.24)	0.37
Shunt infection — no./total no. (%)	5/77 (6)	7/80 (9)	0.73 (0.24–2.21)	0.58

# PREVENZIONE

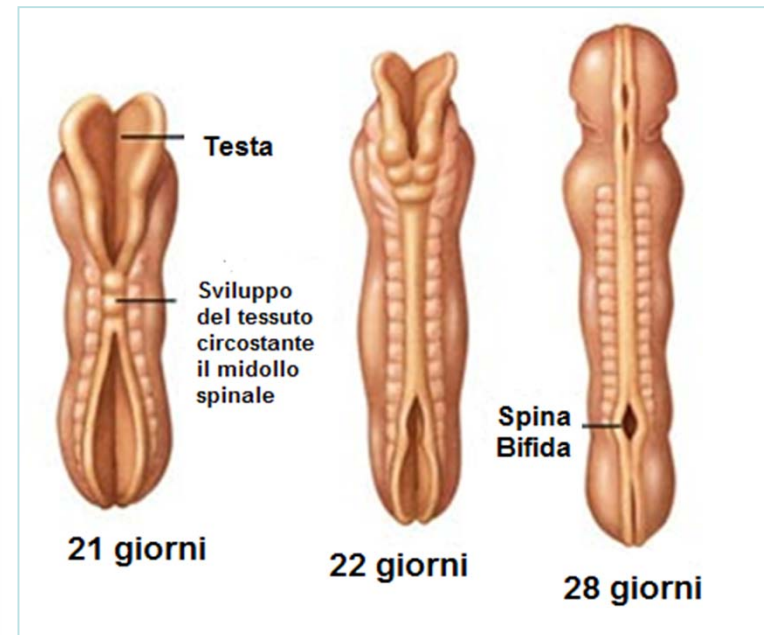
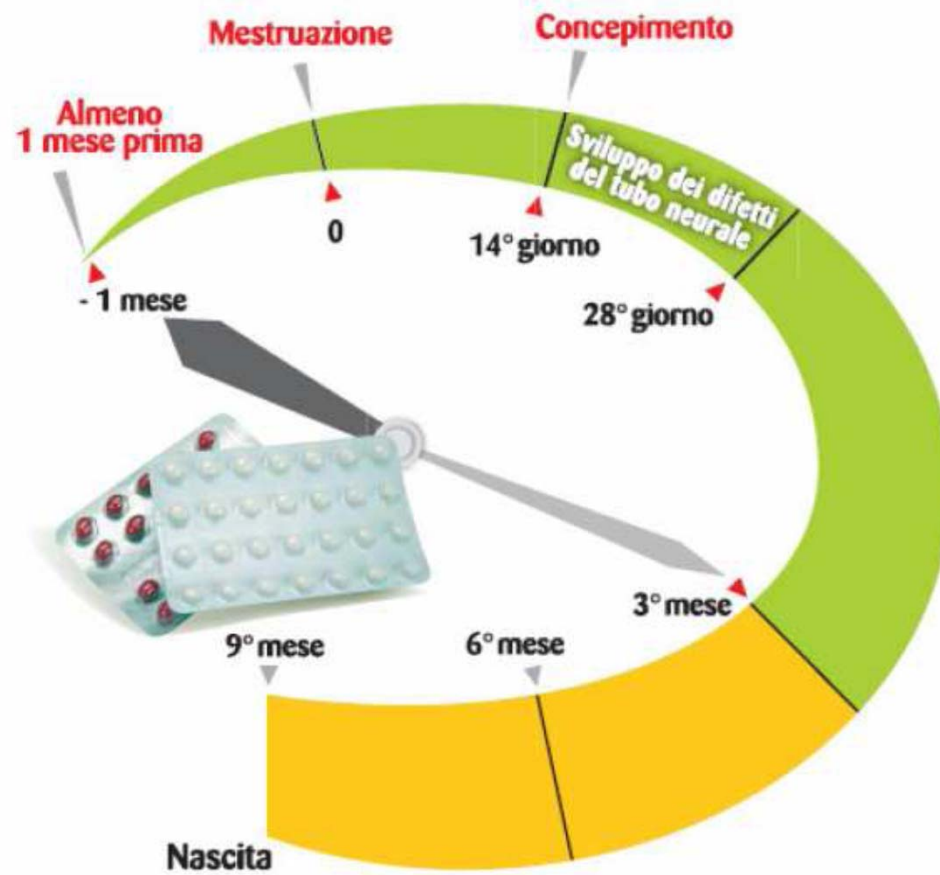


Dieta  
**Supplementazione (0,4 mg/4-5 mg)**  
Fortificazione

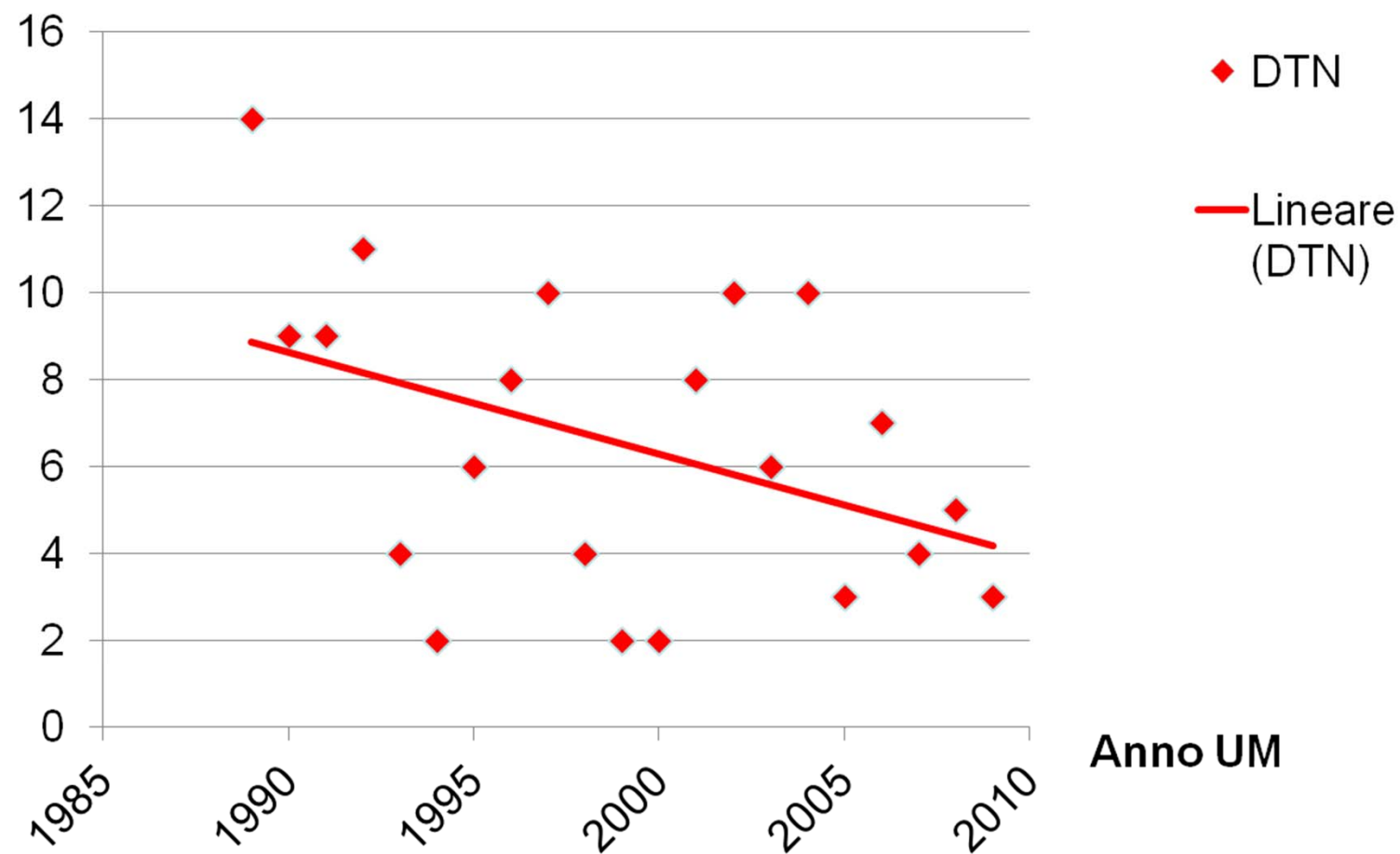


**Acido Folico**  
Un concentrato di protezione  
per il figlio che verrà

## Assunzione dell'**Acido Folico**

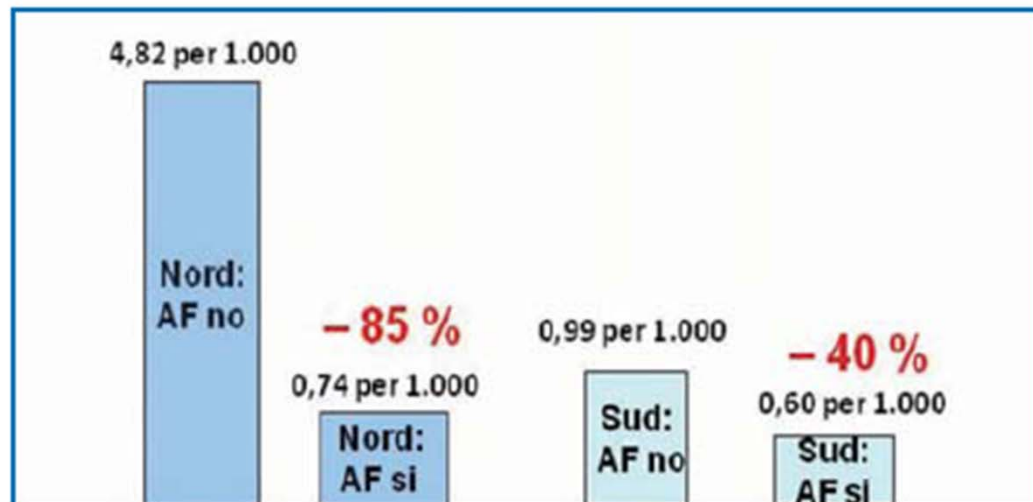


# EPIDEMIOLOGIA





**Decremento dei difetti del tubo neurale ottenuto  
con una supplementazione di acido folico al dosaggio di 0,4 mg/die (AF)  
in uno studio svolto in Cina del Nord e Cina del Sud**





**Prodotti farmaceutici o parafarmaceutici (P) esemplificativi (non esiste lista dei farmaci da banco) utili per prescrivere acido folico**

<b>Prodotto</b>	<b>A F (mg)</b>	<b>N cp</b>	<b>Costo dle</b>	<b>Note</b>
Fertifol	0,4	28	0,09	SSN-A
Folidex	0,4	28	0,09	SSN-A
Folic Acid Biovea	0,8	750	0,05	P-Internet
Multicentrum Materna	0,4	90	0,40	P con altre vitamine
Folac	0,4	60	0,20	P
Serengrav	0,4	40	0,05	P
Folico	0,4	40	0,30	P
Gravigil	0,4	30	0,50	P con altre vitamine*
Azinco gravidanza	0,13	45	0,22	P con altre vitamine*

\*Prodotto in esaurimento **Aggiornato al 06/07/2011**

PAPER

## After-birth abortion: why should the baby live?

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**ABSTRACT**

Abortion is largely accepted even for reasons that do not have anything to do with the fetus' health. By showing that (1) both fetuses and newborns do not have the same moral status as actual persons, (2) the fact that both are potential persons is morally irrelevant and (3) adoption is not always in the best interest of actual people, the authors argue that what we call 'after-birth abortion' (killing a newborn) should be permissible in all the cases where abortion is, including cases where the newborn is not disabled.

**INTRODUCTION**

Severe abnormalities of the fetus and risks for the physical and/or psychological health of the woman are often cited as valid reasons for abortion. Sometimes the two reasons are connected, such as

pathology entails. Many parents would choose to have an abortion if they find out, through genetic prenatal testing, that their fetus is affected by TCS. However, genetic prenatal tests for TCS are usually taken only if there is a family history of the disease. Sometimes, though, the disease is caused by a gene mutation that intervenes in the gametes of a healthy member of the couple. Moreover, tests for TCS are quite expensive and it takes several weeks to get the result. Considering that it is a very rare pathology, we can understand why women are not usually tested for this disorder.

However, such rare and severe pathologies are not the only ones that are likely to remain undetected until delivery; even more common congenital diseases that women are usually tested for could fail to be detected. An examination of 18 European registries reveals that between 2005 and

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A serious philosophical problem arises when the same conditions that would have justified abortion become known after birth. In such cases, we need to assess facts in order to decide whether the same arguments that apply to killing a human fetus can also be consistently applied to killing a newborn human.

Such an issue arises, for example, when an abnormality has not been detected during pregnancy or occurs during delivery. Perinatal asphyxia, for instance, may cause severe brain damage and result in severe mental and/or physical impairments comparable with those for which a woman could request an abortion. Moreover, abnormalities are not always, or cannot always be, diagnosed through prenatal screening even if they have a genetic origin. This is more likely to happen when the disease is not hereditary but is the result of genetic mutations occurring in the gametes of a healthy parent. One example is the case of Treacher-Collins syndrome (TCS), a condition that affects 1 in every 10 000 births causing facial deformity and related physiological failures, in particular potentially life-threatening respiratory

## **ABORTION AND AFTER-BIRTH ABORTION**

Euthanasia in infants has been proposed by philosophers<sup>3</sup> for children with severe abnormalities whose lives can be expected to be not worth living and who are experiencing unbearable suffering.

Also medical professionals have recognised the need for guidelines about cases in which death seems to be in the best interest of the child. In The Netherlands, for instance, the Groningen Protocol (2002) allows to actively terminate the life of 'infants with a hopeless prognosis who experience what parents and medical experts deem to be unbearable suffering'.<sup>4</sup>

Although it is reasonable to predict that living with a very severe condition is against the best interest of the newborn, it is hard to find definitive arguments to the effect that life with certain pathologies is not worth living, even when those pathologies would constitute acceptable reasons for abortion. It might be maintained that 'even allowing for the more optimistic assessments of the potential of Down's syndrome children, this

1. The moral status of an infant is equivalent to that of a fetus, that is, neither can be considered a 'person' in a morally relevant sense.
2. It is not possible to damage a newborn by preventing her from developing the potentiality to become a person in the morally relevant sense.

We are going to justify these two points in the following two sections.

### **THE NEWBORN AND THE FETUS ARE MORALLY EQUIVALENT**

The moral status of an infant is equivalent to that of a fetus in the sense that both lack those properties that justify the attribution of a right to life to an individual.

Both a fetus and a newborn certainly are human beings and potential persons, but neither is a 'person' in the sense of 'subject of a moral right to life'. We take 'person' to mean an individual who is capable of attributing to her own existence some (at least) basic value such that being deprived of this existence represents a loss to her. This means that many non-human animals and mentally retarded human individuals are persons, but that all the individuals who are not in the condition of attributing any value to their own existence are not persons. Merely being human is not in itself a reason for ascribing someone a right to life. Indeed, many humans are not considered subjects of a right to life: spare embryos where research on embryo stem cells is permitted, fetuses where abortion is permitted, criminals where capital punishment is legal.

the moral status of a newborn does not debunk our previous argument. Let us imagine that a woman is pregnant with two identical twins who are affected by genetic disorders. In order to cure one of the embryos the woman is given the option to use the other twin to develop a therapy. If she agrees, she attributes to the first embryo the status of 'future child' and to the other one the status of a mere means to cure the 'future child'. However, the different moral status does not spring from the fact that the first one is a 'person' and the other is not, which would be nonsense, given that they are identical. Rather, the different moral statuses only depends on the particular value the woman projects on them. However, such a projection is exactly what does not occur when a newborn becomes a burden to its family.

### **THE FETUS AND THE NEWBORN ARE POTENTIAL PERSONS**

Although fetuses and newborns are not persons, they are potential persons because they can develop, thanks to their own biological mechanisms, those properties which will make them 'persons' in the sense of 'subjects of a moral right to life': that is, the point at which they will be able to make aims and appreciate their own life.

It might be claimed that someone is harmed because she is prevented from becoming a person capable of appreciating her own being alive. Thus, for example, one might say that we would have been harmed if our mothers had chosen to have an abortion while they were pregnant with us<sup>7</sup> or if they had killed

*La comunicazione ai genitori di una  
patologia prenatale*

- Diagnosi di una patologia prenatale



**Counseling**

## *Cos'è il Counseling*

- Una modalità di lavoro mediante la quale oltre a comunicare ai genitori la diagnosi e la prognosi di una patologia fetale, si tenta di capire cosa essi provano, li si aiuta in un percorso decisionale e si sostengono nelle loro scelte

## *Counseling = Empatia*

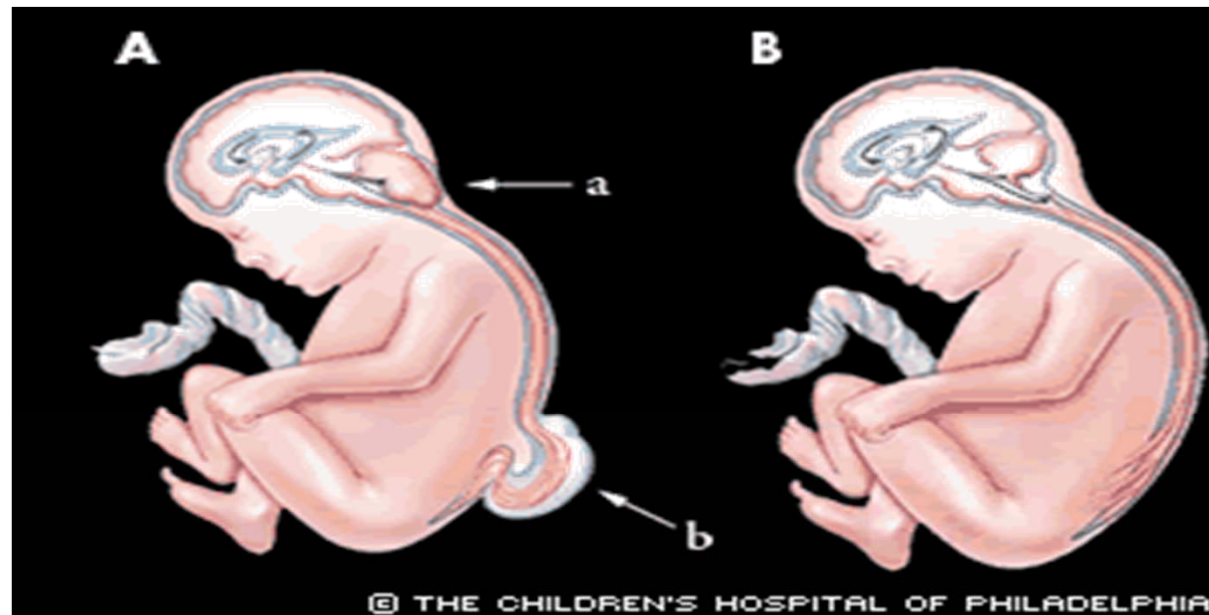
- Immedesimazione con l'io che ho di fronte
- Comprensione delle emozioni e dei bisogni profondi suscitati nell'io dalla circostanza
- Condivisione dell'essere dell'altro non per manipolarlo ma per “aiutarlo”



nella maggior parte dei casi la  
comunicazione di una anomalia  
fetale avviene durante un esame  
ecografico di routine



- Non fare counseling mentre la donna è ancora sdraiata sul lettino
- Trovare uno spazio fisico e “temporale”
- Coinvolgere un collega esperto come aiuto per la spiegazione e per la percezione delle domande
- Comunicare la diagnosi e la prognosi in modo semplice e mediante schemi



Hindbrain herniation before and after myelomeningocele surgery

A. The hindbrain (a) protrudes into the spinal column because of the defect in the lower spine (b).

B. Following early in utero surgical closure of the defect, the hindbrain moves back into a more normal position.

*Esito di 171 bambini con mielomeningocele trattati  
 “aggressivamente” dopo la nascita (Obstet Gynecol 67:1, 1986)*

<b>Livello lesione</b>	<b>Casi con questo livello (%)</b>	<b>Mortalità (%)</b>	<b>QI &gt; 80 (%)</b>	<b>Capacità di camminare (%)</b>	<b>Capacità di camminare senza supporti (%)</b>
<b>Toraco- lombare</b>	<b>37</b>	<b>35</b>	<b>44</b>	<b>71</b>	<b>0</b>
<b>Lombo- sacrale</b>	<b>59</b>	<b>11</b>	<b>65</b>	<b>81</b>	<b>16</b>
<b>Sacrale</b>	<b>4</b>	<b>0</b>	<b>100</b>	<b>100</b>	<b>83</b>

*Associazione tra “segni” ecografici prenatali ed esito  
post-natale in 30 casi (Prenat Diagn 23:311, 2003)*

- 7 bambini “buona prognosi”  
(Motorio e QI normale) -gruppo 1-
- 19 bambini “povera prognosi”  
(Motorio e/o QI anormale) -gruppo 2-

*Associazione tra “segni” ecografici prenatali ed esito  
post-natale in 30 casi      (Prenat Diagn 23:311, 2003)*

	gruppo 1	gruppo 2
Lesione > L4	28%	79%
Ventricolomegalia	57%	94%
Piedi torti	28%	63%

- Dare spazio alle domande
- Offrire test diagnostici di approfondimento (es. ecocardiografia, RM, analisi citogenetica) discutendo le tecniche, i rischi e i benefici. **Pianificare queste indagini in un'altra seduta**
- Lasciare sola la coppia per qualche istante

## *Offrire ulteriori incontri (48-72 ore)*

- Ricontrollo della diagnosi
- Test di approfondimento
- Consulenza specialistica  
(neonatologo/pediatra, chirurgo pediatra,  
neurochirurgo)
- Supporto psicologico



## *Associazioni dei genitori di bambini con malattie congenite*

- Offrire l'opzione di contattare o essere contattati da un gruppo di supporto
- Molte donne che hanno vissuto una simile situazione sono disponibili per il supporto

## *Gruppo di supporto a Monza*

**Segretaria (reperibile 12 ore al giorno)**

2 madri di bambini con S. di Down

2 madri di bambini con Spina Bifida

1 madre di ragazzo con Osteogenesi Imperfetta

1 madre con bambino Idrocefalo

1 madre con bambino con Paralisi Cerebrale Inf.

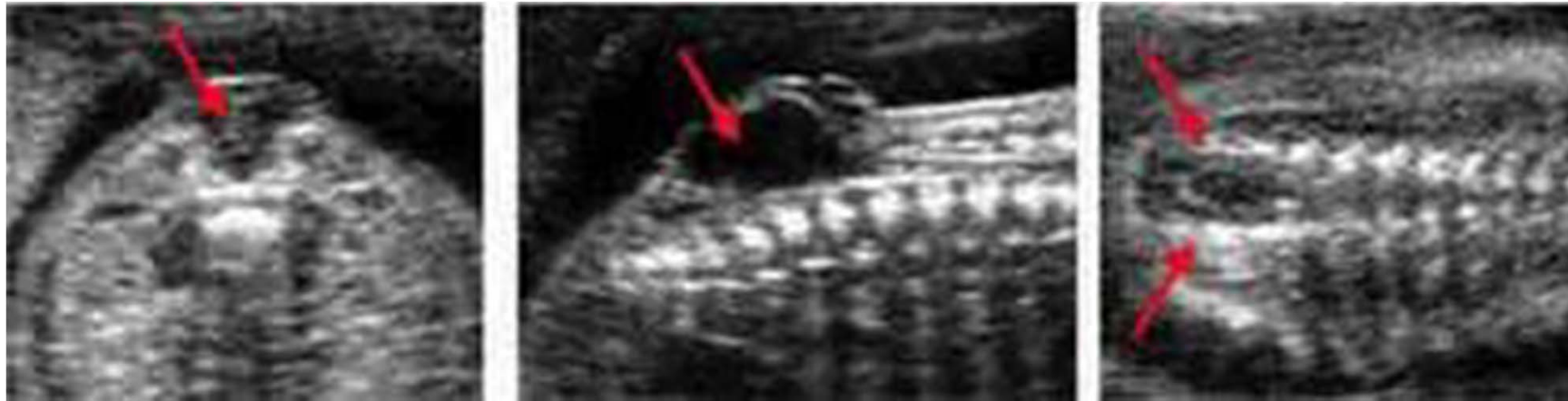
2 madri con bambini con malformazioni facciali

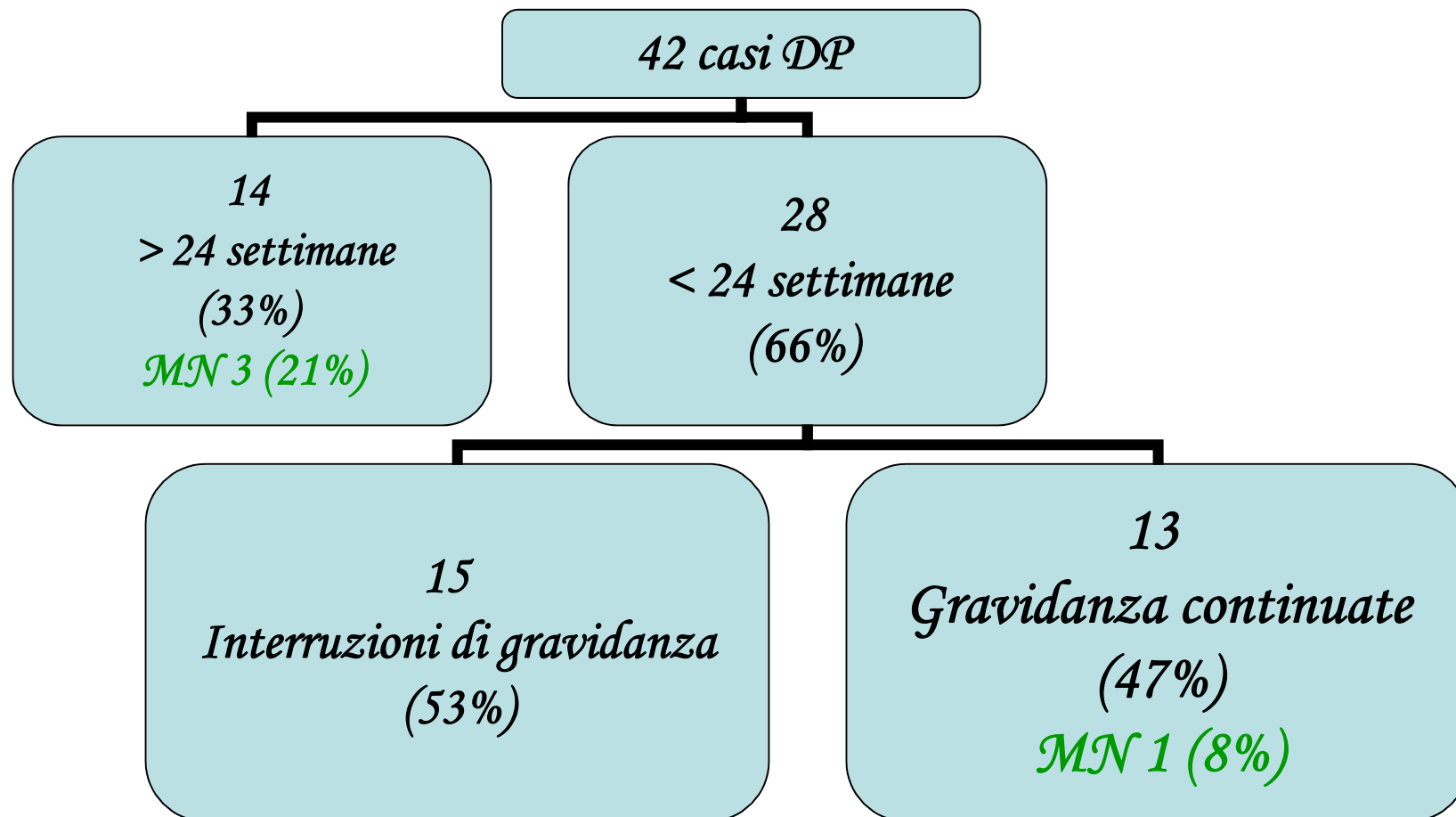
# *Gruppo di supporto di Monza*

## Attività

- Incontri prenatali e postnatali
- Revisione collegiale ogni 6 mesi (con team medici, ostetriche e psicologhe) dei casi incontrati

## *Analisi della nostra casistica*





*47% di gravidanze continuate;  
dato solo di Monza?*

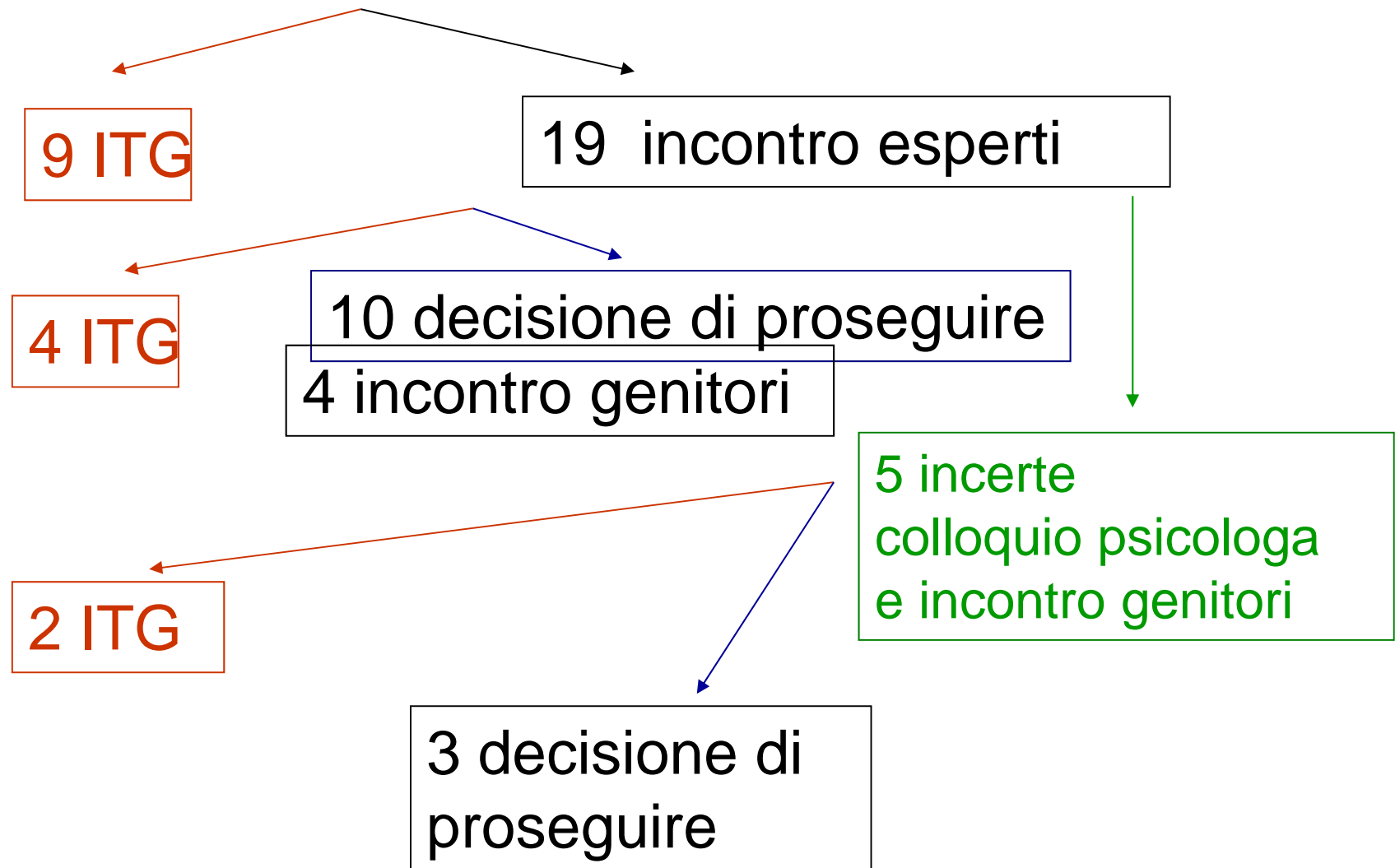
**Mansfield C et al.**

Termination rate after prenatal diagnosis of Down Syndrome, Spina Bifida, Anencephaly, and Turner and Klinefelter Syndromes: a systematic literature review. **Prenat Diagn 19:808, 1999**

**20 studi (EUROCT)**

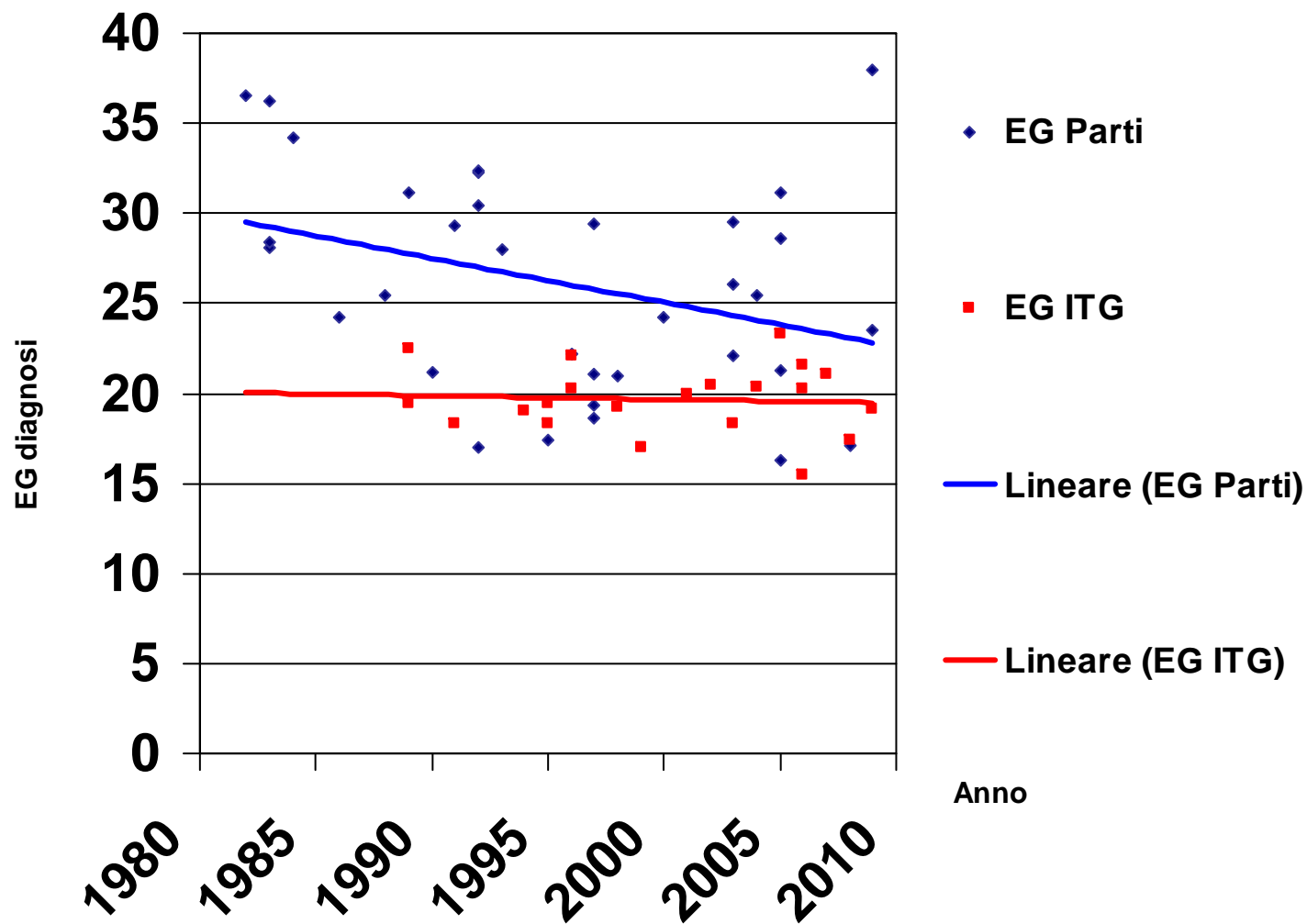
<b>Spina bifida</b>	<b>204 casi diagnosticati</b>
	<b>131 interruzioni di gravidanza (64%)</b>

# 28 casi di Spina Bifida

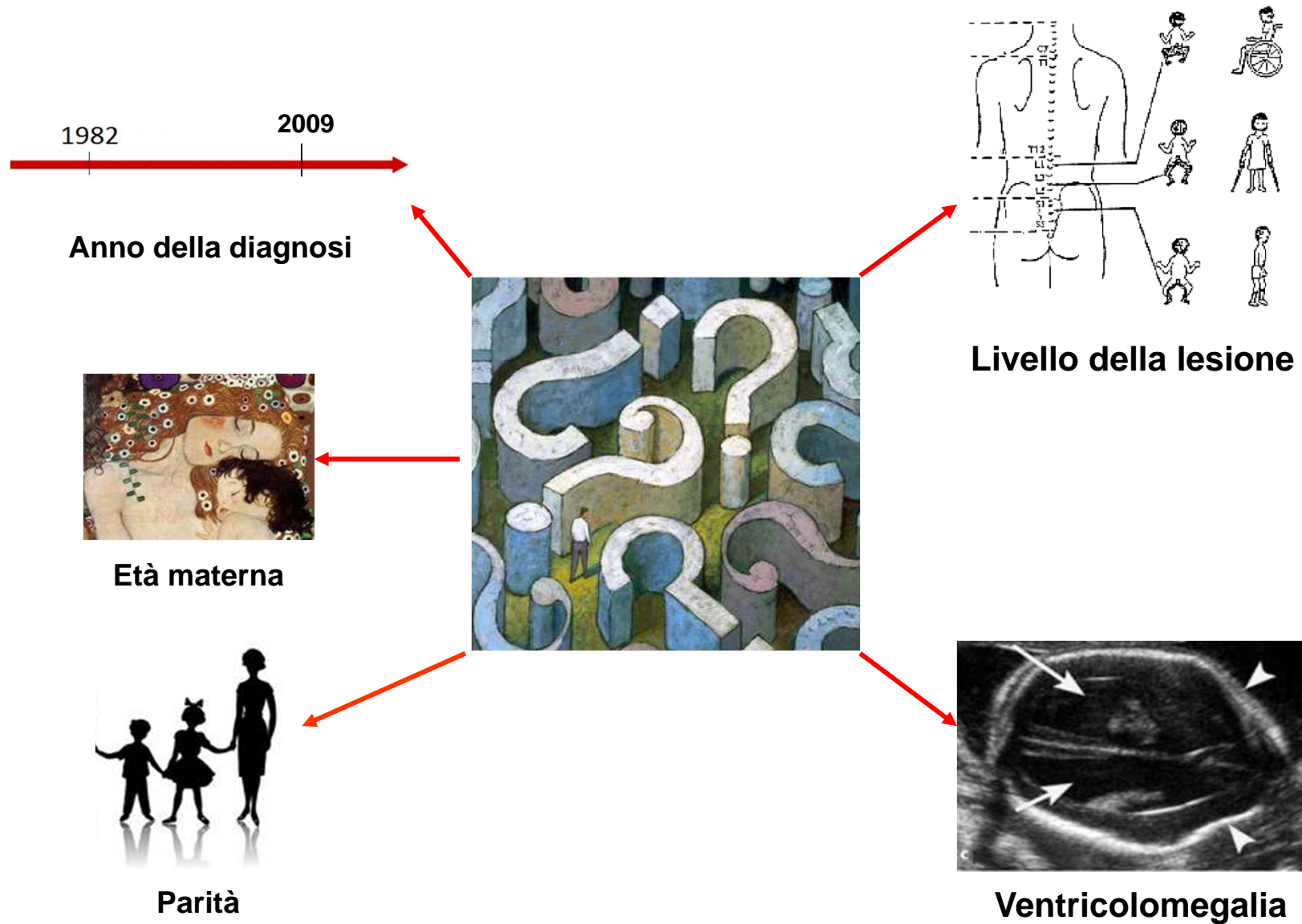




## EG alla diagnosi ITG/PARTI



# Scelta delle donne



*L'uomo diventa  
libero  
solo attraverso  
i legami*

*Franz Kafka*

fare counseling nella diagnosi prenatale  
=  
rispondere ad una circostanza  
non avendo paura di creare legami

# Aumento della sopravvivenza in nati a sempre “più” basse età gestazionali e con basso peso

**Augustines et al.**

Outcome of extremely low-birth-weight  
infants between 500-750 g.

**Am J Obstet Gynecol 2000;182:1113-6**

**Vohr et al.**

Neurodevelopmental and functional outcome  
of extremely LBW infants in the NICH and  
Human Development Neonatal Research  
Network.

**Pediatrics 2000;121:6-26**

Età gestazionale	Vivi	Ritardo psicomotorio
<b>22</b>	<b>0%</b>	
<b>23</b>	<b>19%</b>	
<b>24</b>	<b>32%</b>	<b>66%</b>
<b>25</b>	<b>56%</b>	<b>50%</b>
<b>26</b>	<b>75%</b>	<b>20%</b>
<b>27</b>	<b>100%</b>	<b>20%</b>

Peso neonatale	Bayley MDI < 70	Bayley PDI < 70
<b>&lt; 600 gr</b>	<b>43%</b>	<b>35%</b>
<b>600-800 gr</b>	<b>41%</b>	<b>33%</b>
<b>800-1000gr</b>	<b>30%</b>	<b>26%</b>

# Rinuncia terapeutica ⇒ Eutanasia feto-neonatale

## **Is there a lower limit of viability? Yes**

K. Costeloe (UK): Sessione Plenaria del XVII European Congress of Perinatal Medicine Porto 2000 June 25-28

### **vitale**

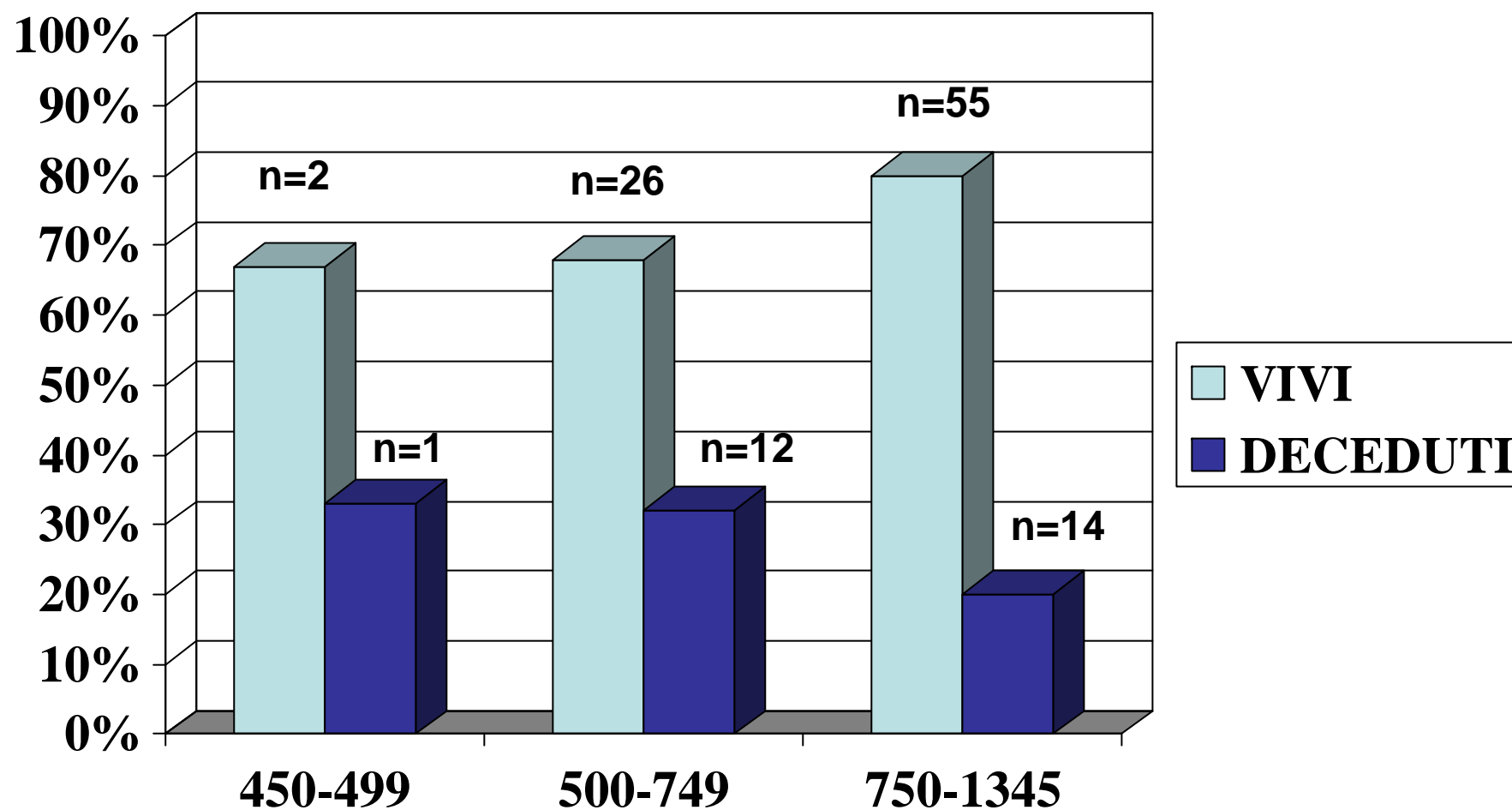
- non un individuo in grado di sopravvivere
- ma un individuo con integrità neuromentale

### **non trattare feto-neonati**

- < 26 settimane (ritardo psicomotorio ⇒ 50%)
- < 800 grammi (MDI <70 ⇒ 42%)

# GENNAIO 2004 - OTTOBRE 2011

## NATI CON PESO TRA 450 E 1345 gr



## Terapia:

Primigravida con preeclampsia severa alla 25 sett. ed iposviluppo fetale, peso previsto ecografico 580 g.

Conduzione “conservativa”- aggressiva: terapia materna e fetale



26.2 sett. ⇒ TC per DIC preclinica ed insufficienza renale materna  
Maschio di 620 g. ⇒ Surfactante, HFO  
Madre guarigione nel post-partum



Per Costeloe (UK)

- sospensione delle cure prenatali
- induzione del parto con prostaglandine

**EUTANASIA**



1 anno



Matteo

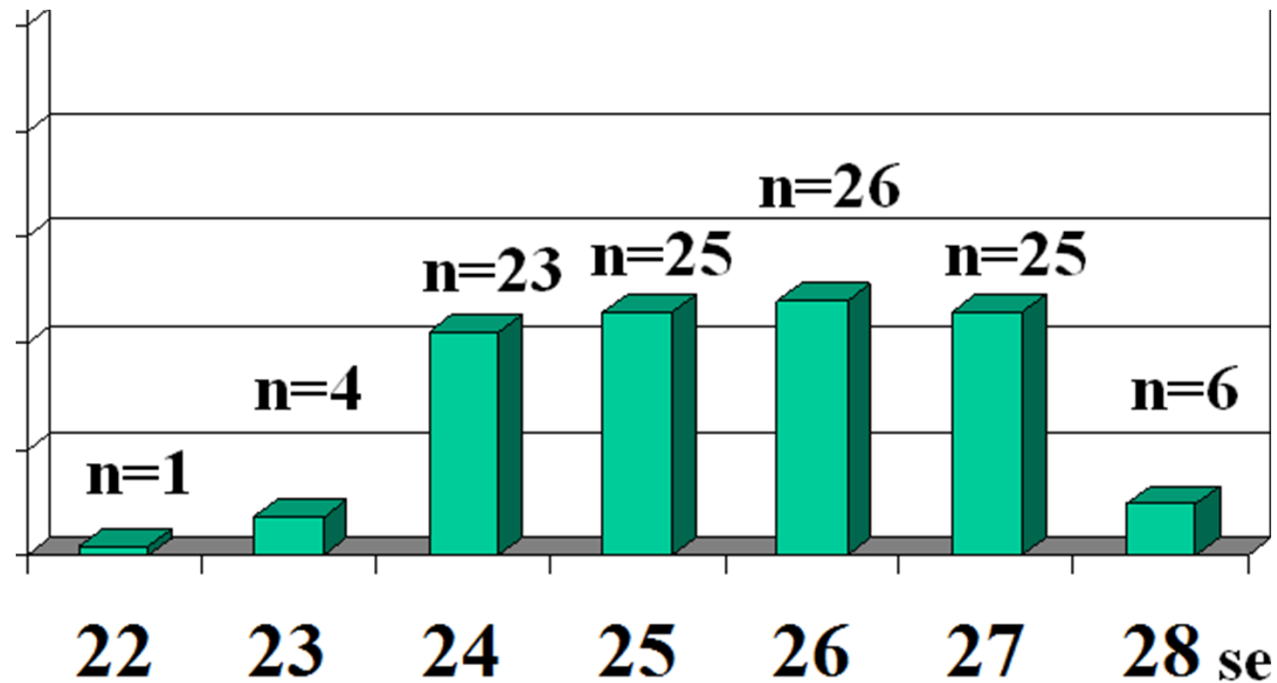
8 anni

**TERAPIA**



# GENNAIO 2004 - OTTOBRE 2011

## NATI TRA 22.0-28.0 SG



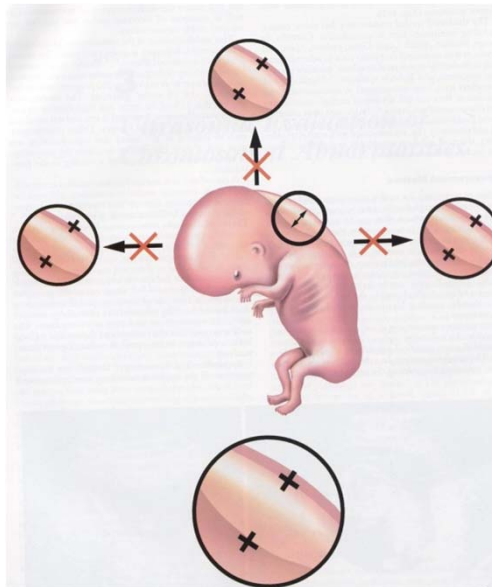
# CONDUZIONE CLINICA

## - 49 PZ CON PP SPONTANEO -

	ATTIVA	NON ATTIVA	P-value
<b>CASI</b>	<b>27 (55%)</b>	<b>22 (45%)</b>	
<i>TRAVAGLIO</i>	23 (85%)	21 (95%)	0.36
<i>TAGLIO CESAREO</i>	11 (41%)	5 (23%)	0.23
<i>EG (settimane gestazionali)</i>	25.2±0.9	24.6±1.0	0.06
<i>PESO (grammi)</i>	762±150	709±111	0.18
<b>VIVI</b>	<b>21 (88%)</b>	<b>8 (36%)</b>	<b>0.00</b>

# **Accesso consapevole alla diagnosi genetica prenatale:**

- test di valutazione del rischio**
- test invasivi**





- Test prenatali per la identificazione della Sindrome di Down

*D. Sicard*

*Presidente Comité National d'Ethique française,  
Le Monde, 2007*

*“È come se con lo screening delle trisomie 18 e 21 (Down) [...] la scienza avesse ceduto alla società il diritto di stabilire che la nascita di certi bambini non è più desiderabile. [In tale contesto] i genitori che ne desiderano la nascita devono esporsi, oltre che al dolore dell'handicap, al rimprovero sociale per non aver accettato la proposta della scienza legittimata dalla legge. In Francia, la diffusione generalizzata dello screening è basata su una proposta, ma nella pratica è divenuta quasi obbligatoria”.*

# Raccomandazioni OMS

## “consumismo prenatale”

*J Med Ethics* 2000;26:444-446 doi:10.1136/jme.26.6.444

### Consumerism in prenatal diagnosis: a challenge for ethical guidelines

Wolfram Henn

 Author Affiliations

#### Abstract

The ethical guidelines for prenatal diagnosis proposed by the World Health Organisation (WHO), as well as by national regulations, only refer to paternity and gender of the fetus as unacceptable, disease-unrelated criteria for prenatal selection, as no other such parameters are at hand so far. This perspective is too narrow because research on complex genetic systems such as cognition and ageing is about to provide clinically applicable tests for genetic constituents of potentially desirable properties such as intelligence or longevity which could be misused as parameters for prenatal diagnosis. Moreover, there is an increasing number of prenatally testable genetic traits, such as heritable deafness, which are generally regarded as pathological but desired by some prospective parents and taken into account as parameters for pro-disability selection. To protect prenatal diagnosis from ethically unacceptable genetic consumerism, guidelines must be clarified as soon as possible and updated towards a worldwide restriction of prenatal genetic testing to immediately disease-determining traits.

Point de vue  
**Dépistage prénatal : Les marchands de risques,**  
par Alexandra Benachi, Roland Gori, Odile Buisson...  
LE MONDE 25.11.09



*Docteur Alexandra Benachi, gynécologue-obstétricienne, MCU-PH, hôpital Antoine-Béclère, Clamart*

*Professeur Roland Gori, psychopathologie, Aix-Marseille*

*Docteur Odile Buisson, échographiste, conseillère ordinaire départementale, Saint-Germain-en-Laye*

*Docteur Marc Althuser, échographiste, Grenoble*

*Docteur Laurent Bidat, échographiste, Saint-Germain-en-Laye*

*Professeur Danièle Brun, psychopathologiste, Paris*

*Professeur Dominique Cabrol, gynécologue-obstétricienne, PU-PH chef de service, hôpital Port-Royal, Paris*

*Professeur Pierre Delion, pédopsychiatre, PU-PH, Lille*

*Docteur Marie-José Del Volgo, MCU-PH, CHU Nord, Marseille*

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*Professeur Jean-Marie Jouannic, gynécologue-obstétricien, PU-PH, hôpital Trousseau, Paris*

*Professeur Christian Laval, sociologue, Paris-X-Nanterre*

*Docteur Sylvain Mimoun, sexologue, Paris*

*Professeur Claire Nihoul-Fékété, chirurgienne pédiatre, PU-PH, Académie de médecine, Paris*

*Professeur Israël Nisand, gynécologue-obstétricien, PU-PH chef de service, Hôpitaux de Strasbourg*

*Serge Portelli, magistrat*

*Docteur Frédéric Prudhomme, président du conseil départemental des Yvelines, Versailles*

*Professeur Jean-François Oury, gynécologue-obstétricien, PU-PH chef de service, hôpital Bernard-Debré, Paris*

*Professeur Georges Vigarello, historien, directeur d'études à l'Ecole des hautes études en sciences sociales*



# Consulenza Genetica

Prax Kinderpsychol Kinderpsychiatr. 2007;56(9):758-71.

## **[Ethical and social aspects of prenatal diagnosis: results from interdisciplinary empirical studies]**

[Article in German]

Irmgard N, Neitzel H.

Frauengesundheitsforschung, Universitätsklinikum Münster. nippert@uni-muenster.de

Since its introduction into maternal health care more than 30 years ago, prenatal diagnosis (PND) is being debated controversially in Germany. The main ethical dilemma associated with PND is the option of selective termination of an affected pregnancy. Another point of concern is PND being presented as a "routine" procedure making it difficult for women to refuse it. When PND was introduced three decades ago there was unanimous agreement that PND should be embedded in pre- and post-test counselling, that PND should only be done with informed consent and that informed consent can only be given if accurate non-biased counselling is provided in a non-directive manner. However, today only a minority of women undergo qualified pre-test (13%) and post-test counselling (18%). Utilization rates of pre- and post-test counselling services are influenced by PND centres and practices and vast regional differences can be observed. Decisions regarding termination of pregnancy depend on many factors including the severity and prognosis of a condition, gestational age and the way in which information about the condition is communicated. In conclusion the uneven availability and accessibility of quality counselling services may impact the wellbeing of women undergoing PND.



# Consulenza Genetica

- **Censimento delle Strutture di Genetica Medica in Italia**  
anno 2007
  - **Istituto Mendel, Roma – Società Italiana di Genetica Umana**  
[www.sigu.net](http://www.sigu.net)
- **560.000 test genetici**
- 311.069 analisi citogenetiche (148.380 postnatali e 162.689 prenatali)
- 227.878 analisi di genetica molecolare (215.551 postnatali e 12.327 prenatali)
- 20.813 analisi immunogenetiche
- **70.154 Consulenze di Genetica Clinica**

# Consulenza genetica: conclusioni

Il numero complessivo delle **Consulenze Genetiche registrate nel 2007 resta basso**, in rapporto alla numerosità dell'attività diagnostica di laboratorio.

Solo 11,5% delle analisi cromosomiche e 13,5% di quelle di genetica molecolare sono state accompagnate dalla Consulenza Genetica.

Si tratta di una percentuale pressoché invariata rispetto a quella del precedente Censimento, che **evidenzia come le raccomandazioni contenute nelle linee guida nazionali e internazionali restino ancora largamente disattese nel nostro Paese.**

# Pericoli per l'autonomia delle donne

Vari studi dimostrano che le donne che si sottopongono a diagnosi genetica prenatale (sia nella forma invasiva che in quella di screening con ecografie mirate o integrate con analisi del sangue materno) raramente hanno piena consapevolezza dei limiti, dei rischi, delle modalità di esecuzione e degli scopi degli screening per la sindrome di Down

- Tymstra T, Bajema C, Beekhuis RJ, Mantingh A. **Women's opinions on the offer and use of prenatal diagnosis.** Prenat Diagn 1991;11:893-898.
- Marteau TM, Cook R, Kid J, Michie S, Johnston M, Slack J, Shaw RW. **The psychological effects of false-positive results in prenatal screening for fetal abnormality: a prospective study.** Prenat Diagn 1992;12:205-14.
- Santalahti P, Latikka AM, Ryyanen M, Hemminki E. **Women's experiences of prenatal serum screening.** Birth 1996;23:101-7.
- Kuppermann M, Nease Jr RF, Learman LA, Gates E, Blumberg B, Washington AE. **Procedure-related miscarriages and Down syndrome-affected births: Implications for prenatal testing based on women's preferences.** Obstet Gynecol 2000;96:511-16.
- Weinans MJ, Huijssoon AM, Tymstra T, Gerrits MC, Beekhuis JR, Mantingh A. **How women deal with the results of serum screening for Down syndrome in the second trimester of pregnancy.** Prenat Diagn 2000;20:705-8.

## **Women's opinions on the offer and use of prenatal diagnosis**

### **Gaps and discrepancies between the women and their health-care providers.**

**Cooley WC, Graham ES, Moeschler JB, Graham JM. Reactions of mothers and medical professionals to a film about Down syndrome. Am J Dis Child 1990;144:1112-1116**

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The **94%** of women believed that the benefits associated with the birth of a DS child outweighed the problems.

Only the **48%** of geneticists thought the same.

The **56%** of geneticists believed the parents should abort a pregnancy with a DS fetus.

Only the **8%** of women agreed with such opinion.

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**Best RG, Sanders AM, Phelan KC, Vincent VA. Declining termination rates for chromosome abnormalities identified prenatally by amniocentesis in southeast United States. Am J Med Genet 1999;A172**

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On 679 consecutive chromosome abnormalities detected at amniocentesis during 11 years (1987-98), including **269** cases of DS, the **32%** of women elected to continue the pregnancy.

**Women's opinions on the offer and use of prenatal diagnosis  
TRASCURATA?**

**Si se alle donne mancano strumenti per un consenso  
informato consapevole**

**Gekas J. Et al.**

**Informed Consent to Serum Screening for Down Syndrome: are  
women given adequate information?**

**Prenat Diagn 1999;19:1-7**

- **Solo il 60% delle donne che hanno ricevuto il consenso informato per lo screening genetico è informato del rischio di aborto dopo amniocentesi.**
- **Il 70% di esse ritengono che il test biochimico non possa avere falsi negativi.**

Retrospective audit of antenatal screening policies for Down's syndrome in eight district general hospital in one health region. (1994-1999)

*Wellesley D et al. BMJ 2002*

Serum screen	Nucal thickening	Maternal age	Detect antenatally	Detect after birth
<b>TOTALE</b> 10%	11%	13%	53%	47%
		< 35aa	35%	65%
		≥ 35aa	67%	33%
			↓	
			23% (43/186)	Refused t.

**Cunningham et al.  
Cost and effectiveness of the California triple marker prenatal  
screening program. Genetics in Medicine 1999;1:200-07**

**Screening mandatorio: 1987-1994 alfa-feto proteina  
1995 -1997 triple test**

**6.031.743 eleggibili**

**3.473.695 (57%) partecipanti (rinunciano 43%)**

**173.740 (5%) positive al test**

**130.496 (75%) DP**

**43.244 (25%) no DP**

**570 feti con Sindrome di Down**

**239 (42%) gravidanze continuate**



**Table 9**  
California Expanded AFP 95-97 decision for elective termination of pregnancy

	Hispanic		White		Asian		Black		Total	
	No.	%	No.	%	No.	%	No.	%	No.	%
Petal Abnormality	DX	TAB	DX	TAB	DX	TAB	DX	TAB	DX	TAB
Anencephaly	157	68.2	92	88.0	20	80.0	10	70.0	297	75.4
Spina bifida	74	60.8	69	72.5	7	57.1	6	83.3	162	67.3
All neural tube defects	259	65.3	195	77.4	33	69.7	18	66.7	532	70.7
Gastroschisis	122	8.2	65	10.8	11	18.2	11	0.0	220	9.1
Omphalocele	19	26.3	16	18.8	2	50.0	3	33.3	45	31.1
All abdominal wall defects	164	18.9	95	21.1	17	35.3	15	13.3	309	21.4
Down syndrome	244	47.5	196	65.8	72	70.8	24	62.5	570	58.6
Trisomy 18	56	55.4	45	73.3	16	68.8	6	50.0	133	66.2
All chromosome abnormalities	443	44.7	401	56.1	138	57.2	60	40.0	1113	51.0
All abnormalities	866	46.1	691	57.6	188	57.4	93	40.9	1954	51.8

DX, prenatally detected; TAB, therapeutic abortion.

## GENETICS

Am J Obstet Gynecol 2008;198:333.e1-333.e8.

## Perceived risk of prenatal diagnostic procedure–related miscarriage and Down syndrome among pregnant women

Aaron B. Caughey, MD, PhD; A. Eugene Washington, MD, MSc; Miriam Kuppermann, PhD, MPH

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**STUDY DESIGN:** We conducted a cross-sectional survey of 1081 English-, Spanish-, or Chinese-speaking women receiving prenatal care in the San Francisco Bay area.

In multivariable linear regression analysis among women younger than age 35 years, the perceived risk of carrying a Down syndrome–affected fetus was higher in women who had not attended college (0.06,  $P$  .019) or had poor self-perceived health status (0.08,  $P$  .045). *Latinas* (0.11,  $P$  .008), women with an annual income less than \$35,000 (0.09,  $P$  .003), and those who had difficulty conceiving (0.09,  $P$  .026) had higher perceived procedure-related miscarriage risk.

## GENETICS

Am J Obstet Gynecol 2008;198:333.e1-333.e8.

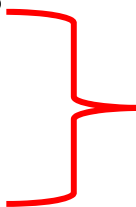
## Perceived risk of prenatal diagnostic procedure–related miscarriage and Down syndrome among pregnant women

Aaron B. Caughey, MD, PhD; A. Eugene Washington, MD, MSc; Miriam Kuppermann, PhD, MPH

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### CONCLUSION:

Women's perceived risks of carrying a Down syndrome–affected fetus or having a procedure-related miscarriage are associated with numerous characteristics that have not been shown to be associated with the actual risks of these events. These perceived risks are associated with prenatal diagnostic test inclination. Understanding patients' risk perceptions and effectively communicating risk is critical to helping patients make informed decisions regarding use of invasive prenatal testing.



# Scelta delle donne

PRENATAL DIAGNOSIS

*Prenat Diagn* 2002; **22**: 769–774.

Published online in Wiley InterScience (www.interscience.wiley.com). DOI: 10.1002/pd.405

## **Factors affecting the decision regarding amniocentesis in women at genetic risk because of age 35 years or older**

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# Scelta delle donne

Dal 1 gennaio 1990 al 31 dicembre 1998, a tutte le donne con età  $\geq 35$  anni è stata offerta la possibilità di effettuare:

- Consulenza genetica individuale
- Ecografia genetica durante il 2° trimestre, definita “anormale” per la presenza di markers di aneuploidia o malformazioni
- E' stato confrontato l'atteggiamento iniziale verso l'amniocentesi (registrato prima della consulenza) con quello finale (dopo l'ecografia)

Table 1—Anamnestic and sociodemographic factors in relation to the *a priori* attitude of women towards amniocentesis [mean  $\pm$  SD or number (%)]

	Decided ( <i>n</i> = 1368)	Undecided ( <i>n</i> = 103)	<i>p</i> value
Maternal age (years)	38.2 $\pm$ 2.0	38.5 $\pm$ 1.7	0.3
Nulliparity	353 (26%)	39 (37%)	0.01
History of $\geq$ 3 spontaneous losses	123 (9%)	9 (9%)	1
Positive genetic history <sup>a</sup>	96 (7%)	4 (4%)	0.3
Remarkable medical history	60 (4%)	5 (5%)	1
High socioeconomic status	246 (18%)	12 (11%)	0.1

<sup>a</sup> For example: family history of malformations, genetic disorders, or mental retardation.

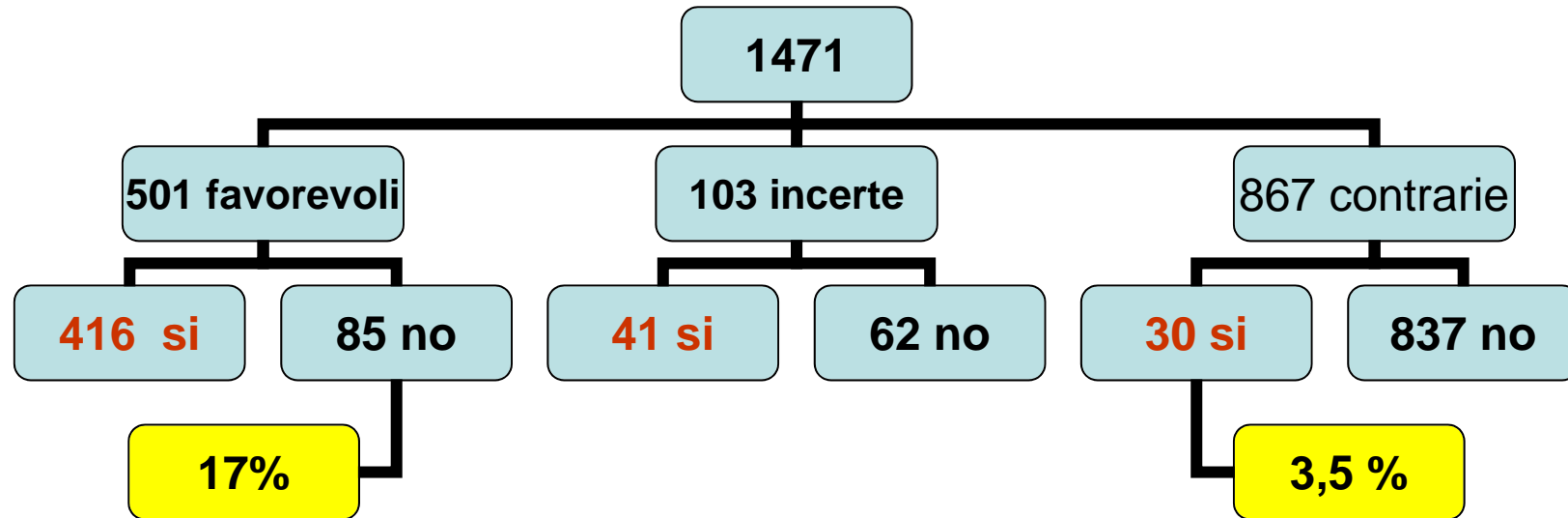
Table 2—Anamnestic and sociodemographic factors in relation to women's *a priori* inclination towards amniocentesis [mean  $\pm$  SD or number (%)]

	<i>A priori</i> Want amniocentesis ( <i>n</i> = 501)	<i>A priori</i> Decline amniocentesis ( <i>n</i> = 867)	<i>p</i> value
Maternal age (years)	38.6 $\pm$ 1.6	37.9 $\pm$ 2.1	<0.001
Nulliparity	136 (27%)	211 (24%)	0.3
Previous recurrent losses	33 (7%)	92 (11%)	0.008
Previous medical history	19 (4%)	41 (5%)	0.5
Positive genetic history <sup>a</sup>	36 (7%)	58 (8%)	0.9
High socioeconomic status <sup>b</sup>	66 (13%)	152 (17%)	0.04

<sup>a</sup> For example: malformations, genetic disorders, mental retardation.

<sup>b</sup> Score 6–8.

# Amniocentesi: opinione “a priori” e scelta



**504 (37%) amniocentesi**  
**34 feti con Sindrome di Down**  
**15 (44%) nati vivi (FN 8)**

# Ecografia e scelta

- Un'ecografia normale modifica più facilmente l'atteggiamento di una donna inizialmente interessata all'amniocentesi di quanto non faccia un'ecografia "anormale" in donne "a priori" non interessate all'amniocentesi (19.6% vs 6.9%, OR 3.2, 95% CI 1.8,3.8)
- Similmente un'ecografia normale ha confermato più facilmente la scelta in una donna "a priori" non interessata all'amniocentesi di quanto un'ecografia "anormale" abbia fatto in donne che inizialmente erano propense ad eseguirla (98% vs 89.5%, OR 5.8, 95% CI 2.5,13.6)



# Diagnosi Prenatale Genetica

(screening sanguigni e/o ecografici e/o procedimenti invasivi)

- Non può mai essere routinaria né proposta sistematicamente, nemmeno nel caso della diagnosi genetica ecografica (per esempio misurazione dello spessore della plica nucale), ma deve essere sempre preceduta da una dettagliata informazione su limiti, rischi, implicazioni e possibilità terapeutiche nell'ambito di una adeguata **consulenza pre-diagnostica** (OMS, 1995)
- Affinché la donna possa compiere una scelta informata ed autenticamente consapevole, conservando la piena libertà di accettare o rifiutare lo screening o il test. Mai devono essere usati termini generici (“piccolo”, “trascurabile”, “grande”) quando si spiega il tasso di rischio, ma vanno forniti dati numerici, nonché il significato di tali dati. Deve essere richiesto chiaramente il consenso informato ed esplicito su numero, tipologia e finalità degli accertamenti.

# Diagnosi Prenatale Genetica

(screening sanguigni e/o ecografici e/o procedimenti invasivi)

- In caso di riscontro di una patologia, la diagnosi prenatale non è da considerarsi terminata (salvo esplicito diniego da parte della donna) senza il coinvolgimento di uno specialista della patologia riscontrata (**consulenza post-diagnostics**), in grado di fornire informazioni sulla patologia, sulla possibilità di un percorso terapeutico e su possibili agevolazioni socio-economiche in grado di assistere la famiglia. Sarà compito del ginecologo diagnosta indirizzare verso tale consulenza specialistica.



Online article and related content  
current as of March 6, 2010.

## Carrier Screening for Gaucher Disease: Lessons for Low-Penetrance, Treatable Diseases

Shachar Zuckerman; Amnon Lahad; Amir Shmueli; et al.

*JAMA*. 2007;298(11):1281-1290 (doi:10.1001/jama.298.11.1281)

**Results** Between January 1, 1995, and March 31, 2003, 10 of 12 Israeli genetic centers (83.3%) offered carrier screening. Carrier frequency was 5.7%, and 83 carrier couples were identified among an estimated 28 893 individuals screened. There were 82 couples at risk for offspring with type 1 GD. Seventy of 82 couples (85%) were at risk for asymptomatic or mildly affected offspring and 12 of 82 couples (15%) were at risk for moderately affected offspring. At postscreening, 65 interviewed couples had 90 pregnancies, and prenatal diagnosis was performed in 68 pregnancies (76%), detecting 16 fetuses with GD (24%). Pregnancies were terminated in 2 of 13 fetuses (15%) predicted to be asymptomatic or mildly affected and 2 of 3 fetuses (67%) with predicted moderate disease. There were significantly fewer pregnancy terminations in couples who in addition to genetic counseling had medical counseling with a GD expert (1 of 13 [8%] vs 3 of 3 with no medical counseling [100%],  $P = .007$ ).

# Uso routinario test rischio di una “non scelta”

- Garantire la **libertà nella scelta**
- **Integrare sistematicamente la diagnostica prenatale con una “consulenza pre-diagnostica” e una “post-diagnostica”**
- La diagnosi genetica prenatale non è eticamente neutra: come tutti gli atti umani è una scelta
- Le scelte richiedono una reale conoscenza dei dati e implicano una responsabilità.
- L'autonomia delle donne nelle decisioni sulla loro gravidanza può essere seriamente compromessa da un uso routinario

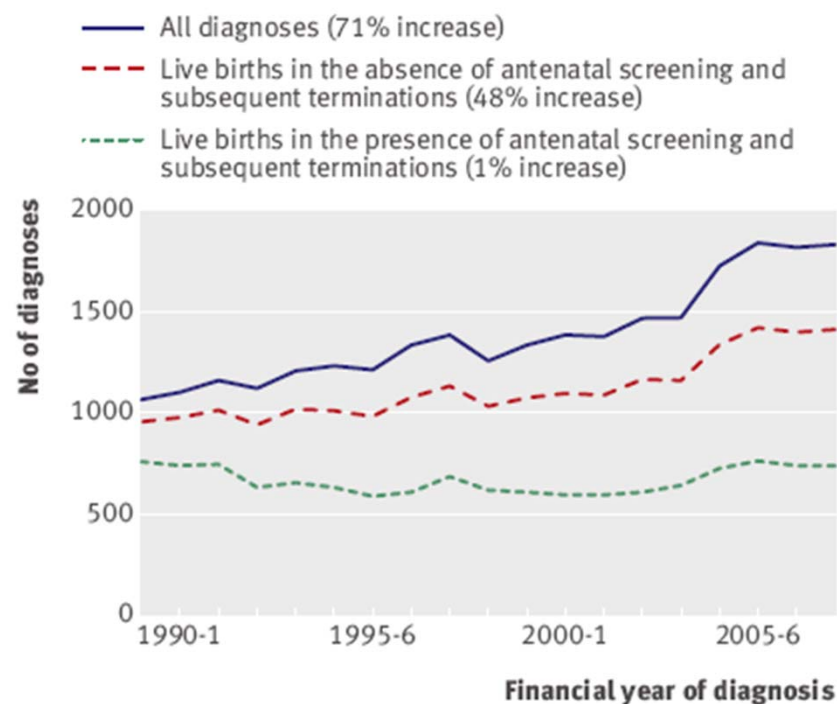
## Trends in Down's syndrome live births and antenatal diagnoses in England and Wales from 1989 to 2008: analysis of data from the National Down Syndrome Cytogenetic Register

Joan K Morris, professor of medical statistics, Eva Alberman, emeritus professor

Wolfson Institute of Preventive Medicine, Barts and the London School of Medicine and Dentistry, Queen Mary University of London, London EC1M 6BQ

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Cite this as: *BMJ* 2009;339:b3794  
doi:10.1136/bmj.b3794



**Fig 1** | Down's syndrome diagnoses and live births according to year of diagnosis and presence or absence of antenatal screening and subsequent terminations

# Trends in Down's syndrome live births and antenatal diagnoses in England and Wales from 1989 to 2008: analysis of data from the National Down Syndrome Cytogenetic Register

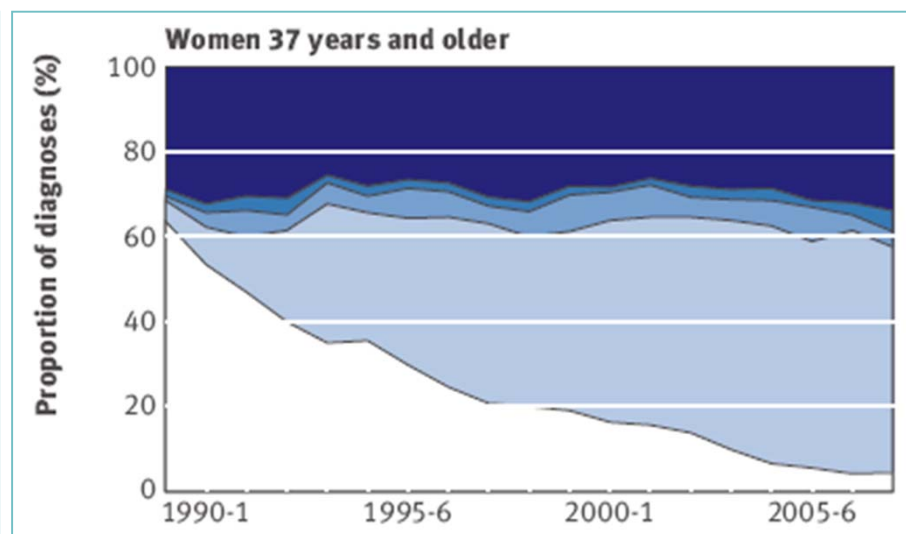
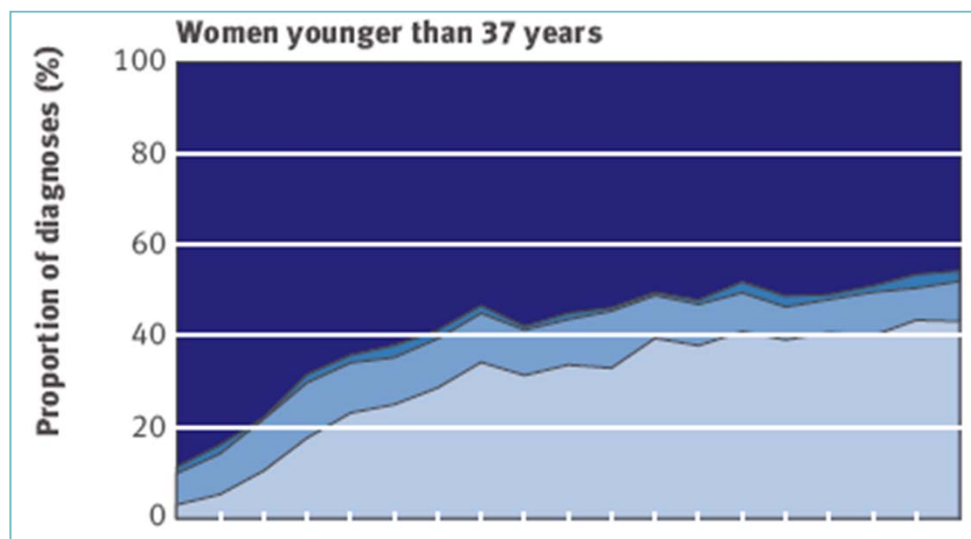
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Cite this as: *BMJ* 2009;339:b3794  
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■ Postnatal      ■ Ultrasound after 16 weeks      □ Maternal age  
■ Other/missing      ■ Screening







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[DSA Response to the British Medical Journal research on Down's syndrome births - released 27th October](#)



## DSA response statement:

Carol Boys, Chief Executive for Down's Syndrome Association said, "We realise that tests will continue to become more accurate at increasingly earlier stages of pregnancy. It is therefore even more important that families undergoing the screening process are given non-directive counselling and accurate, up-to-date information about Downs' syndrome."

Attitudes towards people with Down's syndrome have changed dramatically over the last 20 years or so. Children with Down's syndrome are being educated in mainstream schools, and adults are working in local employment and living semi-independently.

## **“STORIA NATURALE” TRASCURATA ???**

1. Tanto più precoce è un test diagnostico, tanti più feti con aneuploidie vengono diagnosticati.
2. Feti che vengono “persi” naturalmente diventano oggetto di scelta decisionale.
3. Nessuno studio per verificare se i feti spontaneamente abortiti hanno maggior positività ai test precoci.



***Macintosh MC. Br J Obstet Gynaecol 1995***

Selective miscarriage of Down's syndrome fetuses in women aged 35 years and old.

- Il 20% di feti con Sindrome di Down sono abortiti spontaneamente tra la 10<sup>a</sup> e 16<sup>a</sup> settimana
- Il 45% tra il concepimento ed il termine della gravidanza.

# **“COSTO EMOTIVO” TRASCURATO???**

Le donne che ricevono un risultato “falso positivo” riportano un sentimento di depressione e ansia per tutto il resto della gravidanza.

1. Sentimenti di tradimento nei confronti del proprio bambino
2. Denunciano di essere state forzate ad interrompere il rapporto affettivo col bambino nell'attesa del risultato del cariotipo
3. Anche in presenza di cariotipo normale temono che il test positivo possa rappresentare un fattore di rischio per il bambino

## **ACCETTABILITA' SOCIALE CAMBIATA DALLO SCREENING???**

- L'introduzione dei test di screening condiziona la soglia di accettabilità della Sindrome di Down?
- La nascita di un bambino affetto sarà ritenuto un “caso mancato”?

# Gruppo-choc su Facebook contro down, scoperto autore

E' un diciannovenne cingalese. L'indagine partita da Catania

06 marzo, 19:31

◀ Indietro | 🖨 Stampa | ✉ Invia | ✎ Scrivi alla redazione | 💬 Suggestisci ()

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CATANIA - Una bravata informatica di un cingalese di 19 anni, da tempo sotto cure psichiatriche, che aveva come scopo una gara per ottenere più contatti possibile sul social network di Internet. E' quanto c'era dietro al gruppo-choc apparso nel febbraio scorso su Facebook intitolato "Giochiamo al tiro al bersaglio con i bambini down: è l'unica fine che meritano questi parassiti", che aveva come 'logo' la foto di un neonato disabile con la parola 'scemo' scritta sulla fronte e che in poche poche ore era riuscito a raccogliere oltre 1.300 iscritti.