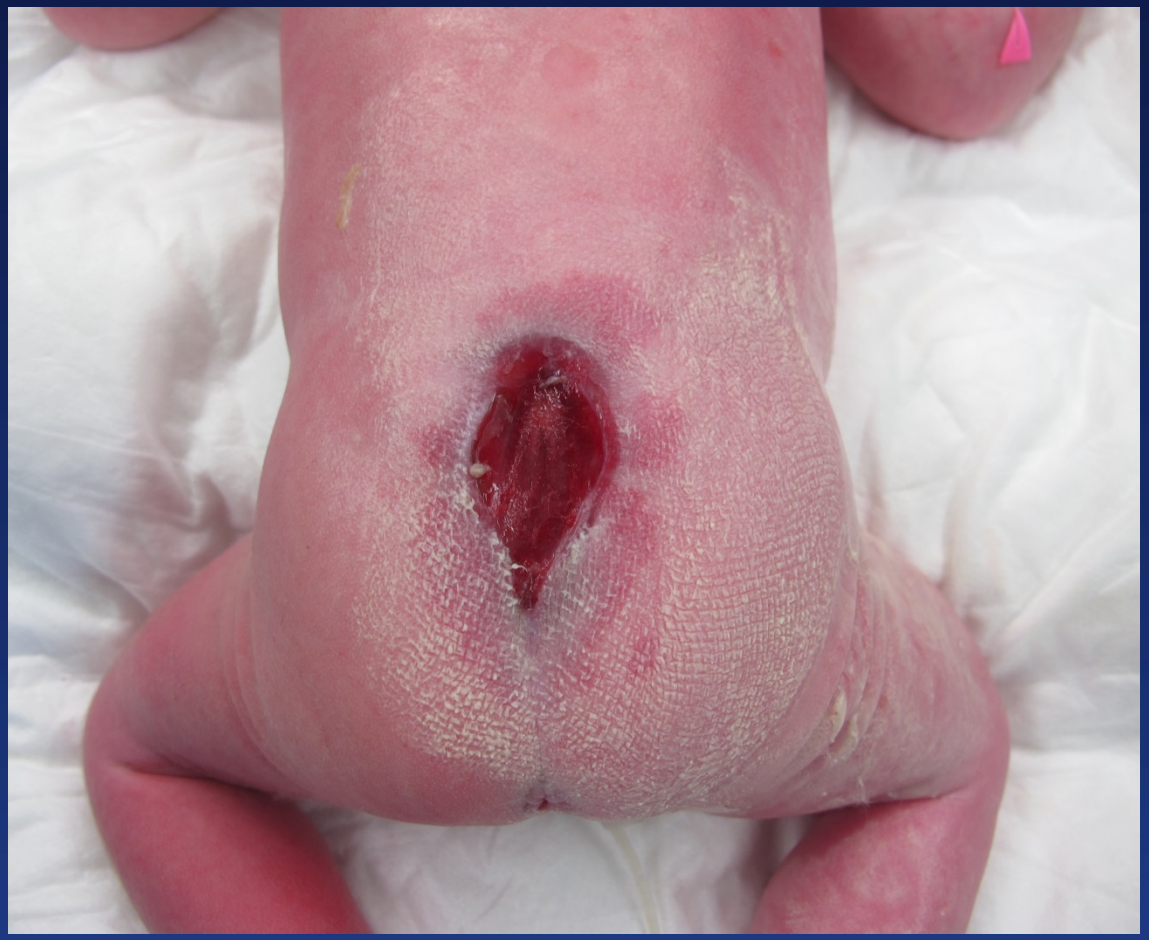
Pränatal-Medizin München

Prenatal MMC: Diagnosis and treatment options in Germany

Karl-Philipp Gloning, Munich, Germany

European Club for Fetal Surgery
January 14 – 16, 2015; Sils, Switzerland

Neural tube defect – unknown until birth



Baby born
August, 24th 2013

Defect not known before birth

No diagnosis
No treatment option

NTD-Screening: intracranial translucency

Ultrasound Obstet Gynecol 2010; 35: 133–138
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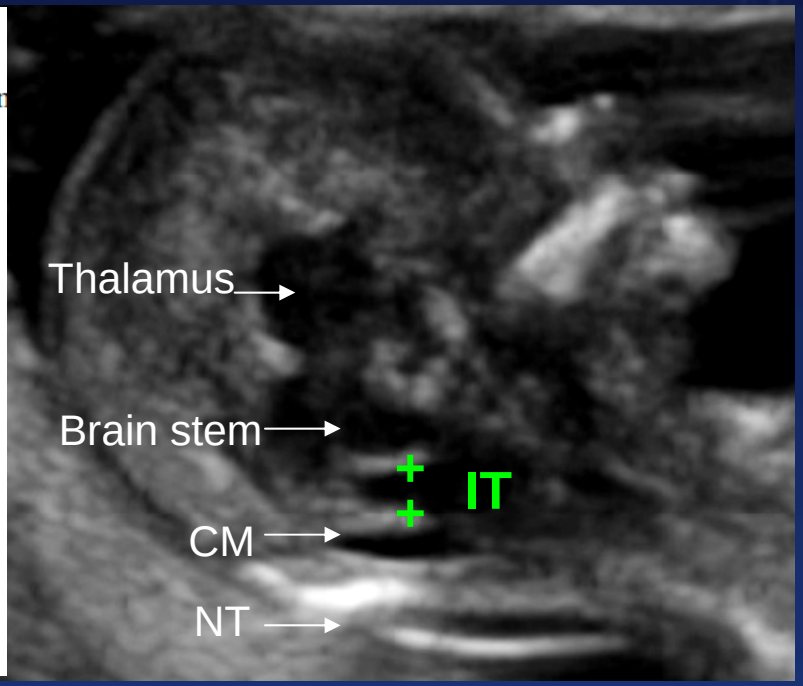
Editorial

From nuchal translucency to intracranial translucency: towards the early detection of spina bifida

R. CHAOUI†*and K. H. NICOLAIDES‡



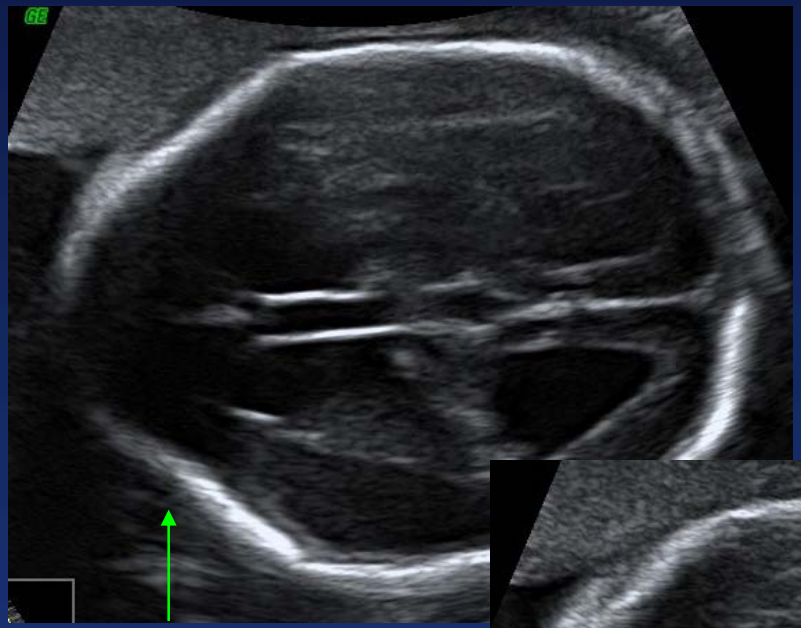
Intracranial translucency:

Posterior brain stem and chorioid plexus of IV. ventricel

- not visible in cases of spina bifida / MMC

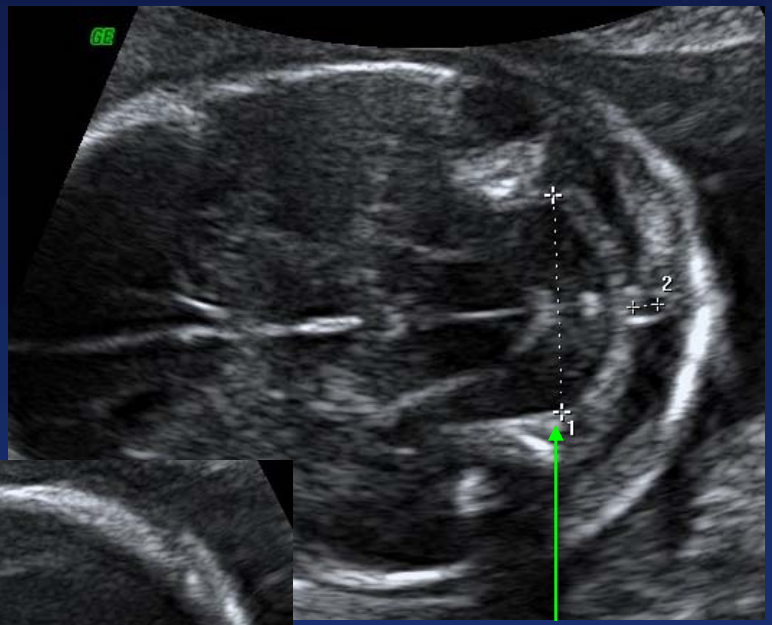
Clinical use and significance unknown (overestimated)

NTD-Screening 20 weeks: Lemon-sign, Banana-sign, ventriculomegaly

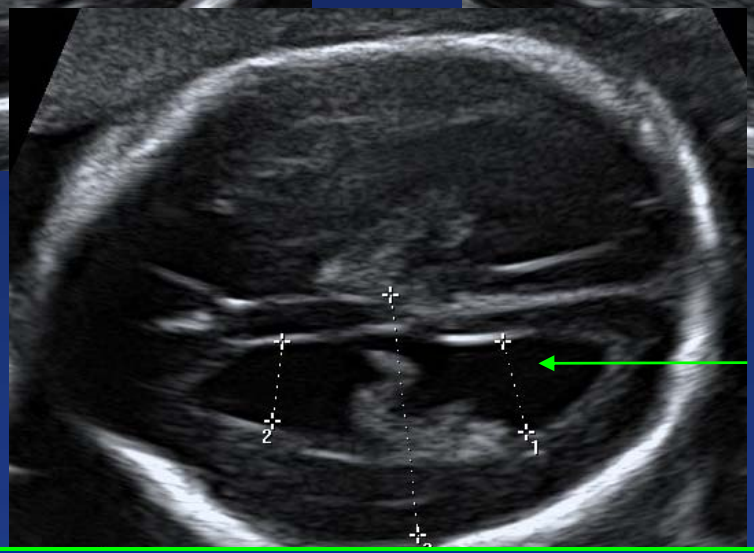


Lemon-Sign

20+6



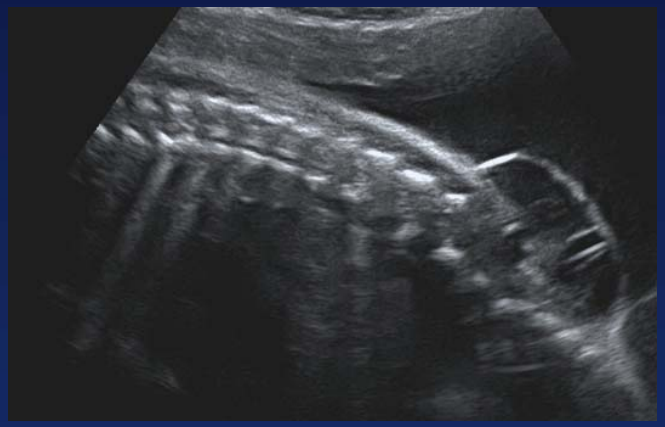
Banana-Sign

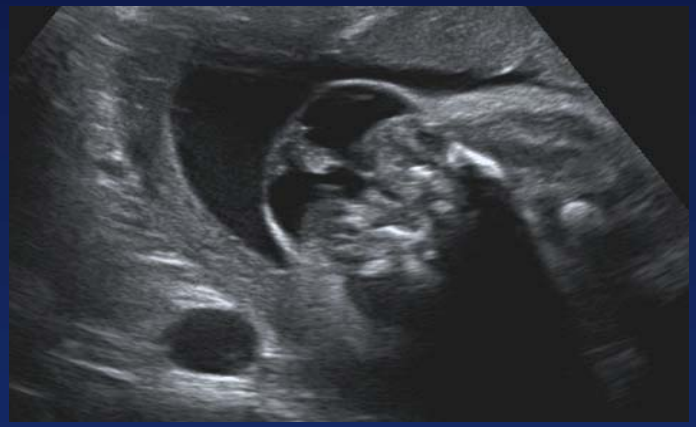


ventriculomegalie

- The diagnosis is made by brain and skull signs

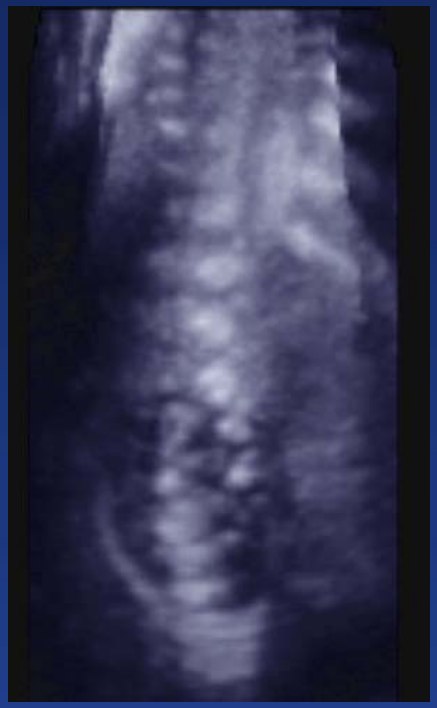
Spina bifida aperta lumbosacralis and MMC vertebral defect and skin over defect

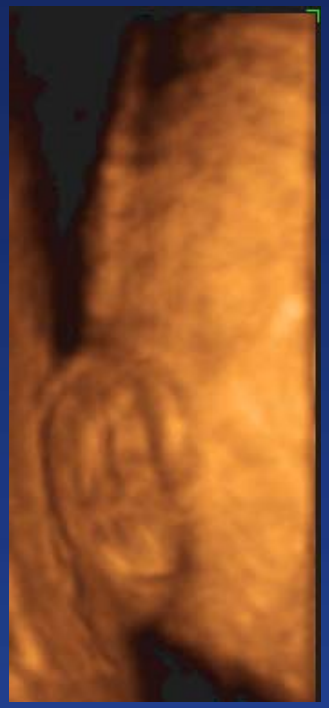


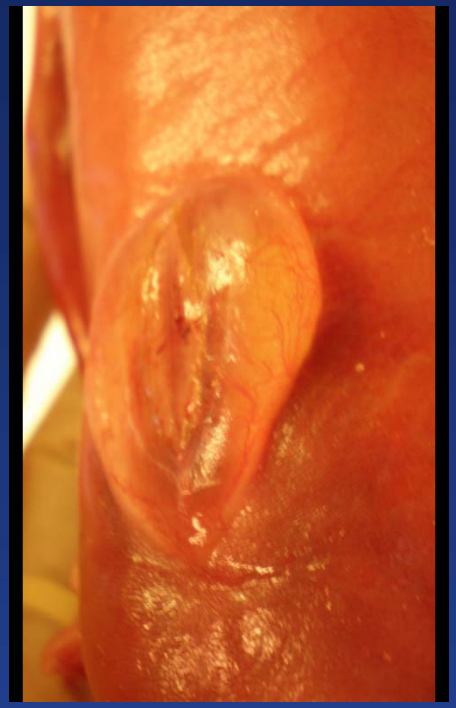


21+6

3D-Surface







after termination

MMC: Prevalence?

- Germany:
- No national registry for fetal / neonatal malformations
- Registry for Mainz and Saxony-Anhalt only („Eurocat“)
- Birth prevalence for MMC unknown

- Pränatal-Medizin München: N = 264 (1994 – 2014)
- ~ 13 cases / year (diagnosed mainly during 2nd trimester)
- ~ 4 cases / year lifeborn and operated
- > 2/3 are terminated, IUD, late termination, stillbirth
- (over all 11 % with chromosomal aberration)

MMC: Prevalence?

- Pränatal-Medizin München: N = 264 (1994 – 2014)
- ~ 13 cases / year (diagnosed mainly during 2nd trimester)
- ~ 4 cases / year lifeborn and operated

- Population of referals about 15.000 to 20.000 pregnancies
- In Bavaria ~ 100.000 newborns / year
- → about 25 with MMC born alive in Bavaria

- In Germany ~ 600.000 newborns / year
- → about 150 with MMC born alive in Germany

MMC: Prevalence

EUROCAT Prevalence Data Tables

A5 - Spina Bifida (per 10,000 births) for the following registries: All Registries, from 2008 - 2012

	2008	2009	2010	2011	2012	Totals
Full Member Registries						
Mainz (Germany						
Total Cases	2	5	1	1	7	16
Live Births	1	4	0	1	2	8
Fetal Deaths	0	0	0	0	0	0
Terminations	1	1	1	0	5	8
Population	3293	3160	3168	3149	3246	16016
Prevalence	6.07	15.82	3.16	3.18	21.57	9.99
Total Cases exc. chromosoma	1	5	1	1	7	15
Prevalence exc. chromosoma	3.04	15.82	3.16	3.18	21.57	9.37
Saxony-Anhalt (Germany)						
Total Cases	16	14	7	2	12	51
Live Births	5	4	4	2	1	16
Fetal Deaths	0	0	0	0	0	0
Terminations	11	10	3	0	11	35
Population	17763	17213	17363	16906	16951	86196
Prevalence	9.01	8.13	4.03	1.18	7.08	5.92
Total Cases exc. chromosoma	16	14	7	2	11	50
Prevalence exc. chromosoma	9.01	8.13	4.03	1.18	6.49	5.80

Pränatal-Medizin München

Total cases	18	8	11	11	8	**56**
Live birth	5	1	4 (1 Patch)	4	2 (1 Patch)	**16** **28,5 %**
Terminations	13 (3)	7 (3)	7 (3)	7 (3)	6 (4)	**40 (16)** **71,5 %**
Chrom. Aberration	3	1	1	3	2	**10** **17,8 %**
Prevalence (estimated population 15.000 per year)						~ 8 in 10.000

MMC: Treatment options

Percutaneous minimally invasive fetoscopic surgery

Deutschen Zentrums für Fetalchirurgie & minimal-invasive Therapie (DZFT)
Prof. Thomas Kohl

Ultrasound Obstet Gynecol 2014; **44**: 515–524
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Percutaneous minimally invasive fetoscopic surgery for spina bifida aperta. Part I: surgical technique and perioperative outcome

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KEYWORDS: Chiari-II malformation; fetal surgery; fetoscopy; fetus; hydrocephalus; spina bifida

ABSTRACT

Objectives *To analyze the current technical approach of percutaneous minimal-access fetoscopic closure of spina bifida aperta (SBA) and provide an overview of its development in ovine and human fetuses.*

'Management of Myelomeningocele Study' (MOMS) provided level-I evidence that prenatal SBA closure may preserve leg function and reduce the severity of hindbrain herniation and hydrocephalus in affected fetuses[1]. Yet, the open surgical approach is associated with significant maternal morbidity, as it requires maternal laparotomy

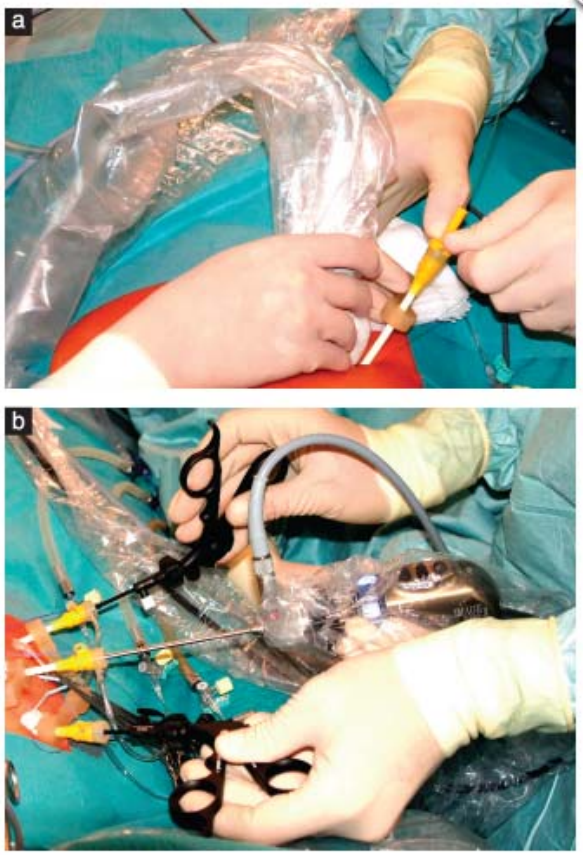


Figure 1. Percutaneous minimal-access fetoscopic surgery for spina bifida aperta: external aspect of the set-up. The procedure is performed via three (or four) trocars, each with an external diameter of 5 mm, that are placed into the amniotic cavity under continuous ultrasound monitoring by a Seldinger approach (a). Following partial amniotic carbon dioxide insufflation and fetal posturing, surgical dissection of the malformation is carried out using fetoscopic instruments (b).

51 cases operated between July 2010 and June 2013:
~ 17 cases / year

MMC: Treatment options

Percutaneous minimally invasive fetoscopic surgery

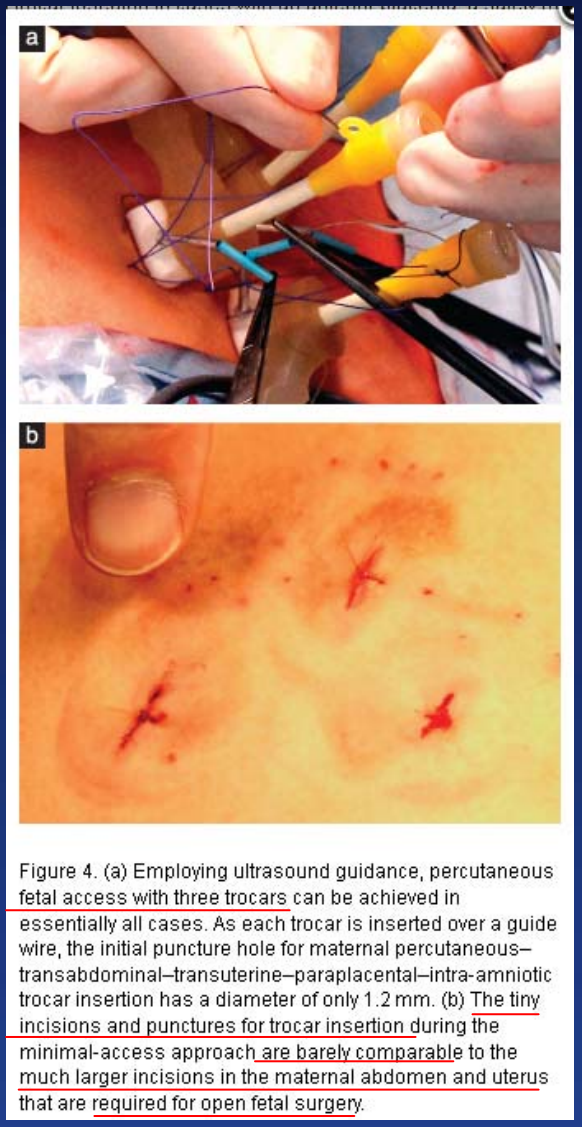
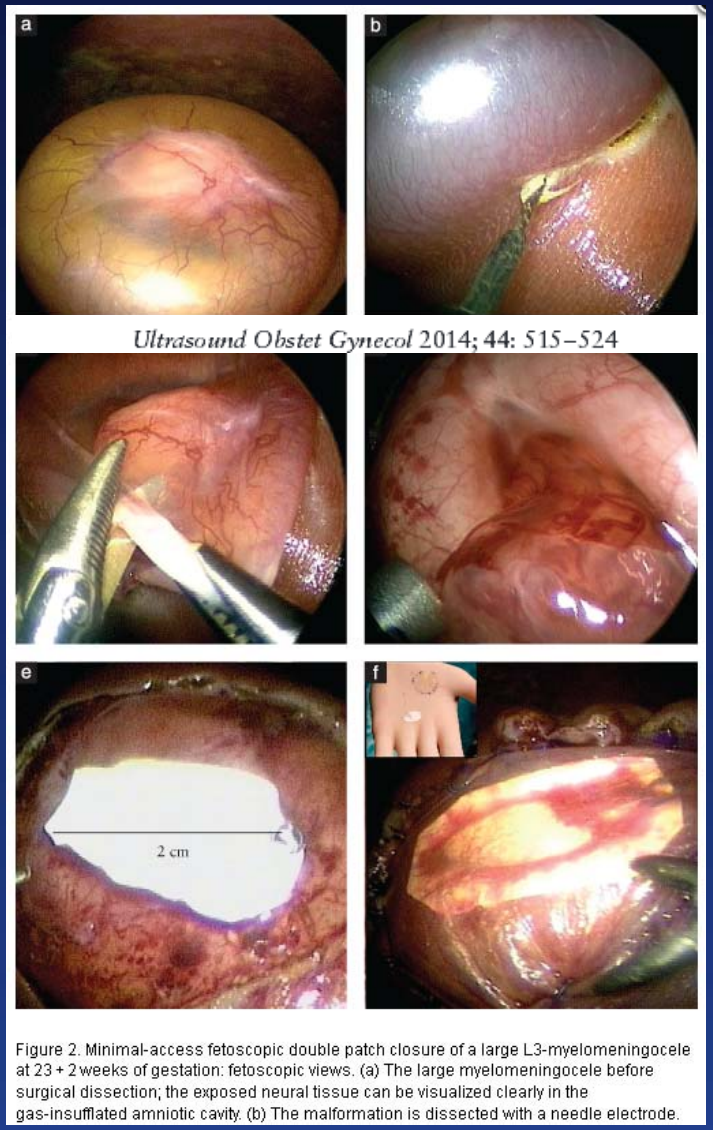


Figure 4. (a) Employing ultrasound guidance, percutaneous fetal access with three trocars can be achieved in essentially all cases. As each trocar is inserted over a guide wire, the initial puncture hole for maternal percutaneous–transabdominal–transuterine–paraplacental–intra-amniotic trocar insertion has a diameter of only 1.2 mm. (b) The tiny incisions and punctures for trocar insertion during the minimal-access approach are barely comparable to the much larger incisions in the maternal abdomen and uterus that are required for open fetal surgery.



Ultrasound Obstet Gynecol 2014; 44: 515–524

Figure 2. Minimal-access fetoscopic double patch closure of a large L3-myelomeningocele at 23 + 2 weeks of gestation: fetoscopic views. (a) The large myelomeningocele before surgical dissection; the exposed neural tissue can be visualized clearly in the gas-insufflated amniotic cavity. (b) The malformation is dissected with a needle electrode.

MMC: Treatment options

Percutaneous minimally invasive fetoscopic surgery

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Opinion

Percutaneous minimal-access fetoscopic surgery for myelomeningocele – not so minimal!

the deleterious effects of amniotic fluid on exposed neural tissue and prevent cerebrospinal fluid leakage through the defect to reverse hindbrain herniation. There are no data on reversal of hindbrain herniation from which to draw and there is the disturbing result of two postnatal deaths from severe brain-stem dysfunction attributed to Chiari-II malformation – a phenomenon not observed after open fetal surgery, in which some degree of reversal of the Chiari malformation is seen almost universally. Adding to this concern, there is usually no skin closure performed, resulting in an exposed, non-epithelialized patch at birth.

If Professor Kohl truly expects this approach to gain acceptance over open fetal surgery, which has been subjected to the most rigorous of outcome analysis, it is time for the data on surgical and neurological assessment of this cohort – which I believe to be currently available – to be published. This should include documentation of the status of the hindbrain, rate of ventriculoperitoneal shunting and clear criteria by which it was performed, anatomical *vs* observed level of neurological function, ambulatory status and, ultimately, bowel, bladder and cognitive function. Most importantly, this should be reported by third-party observers to remove the potential for investigator bias.

The Management of Myelomeningocele Study avoidance of hysterotomy, and improved maternal cosmesis to their patients, they are obligated to demonstrate rigorously at least equal postnatal neurological outcomes to those achieved after open fetal surgical repair. Until they do, this is best viewed as an ongoing example of human experimentation.

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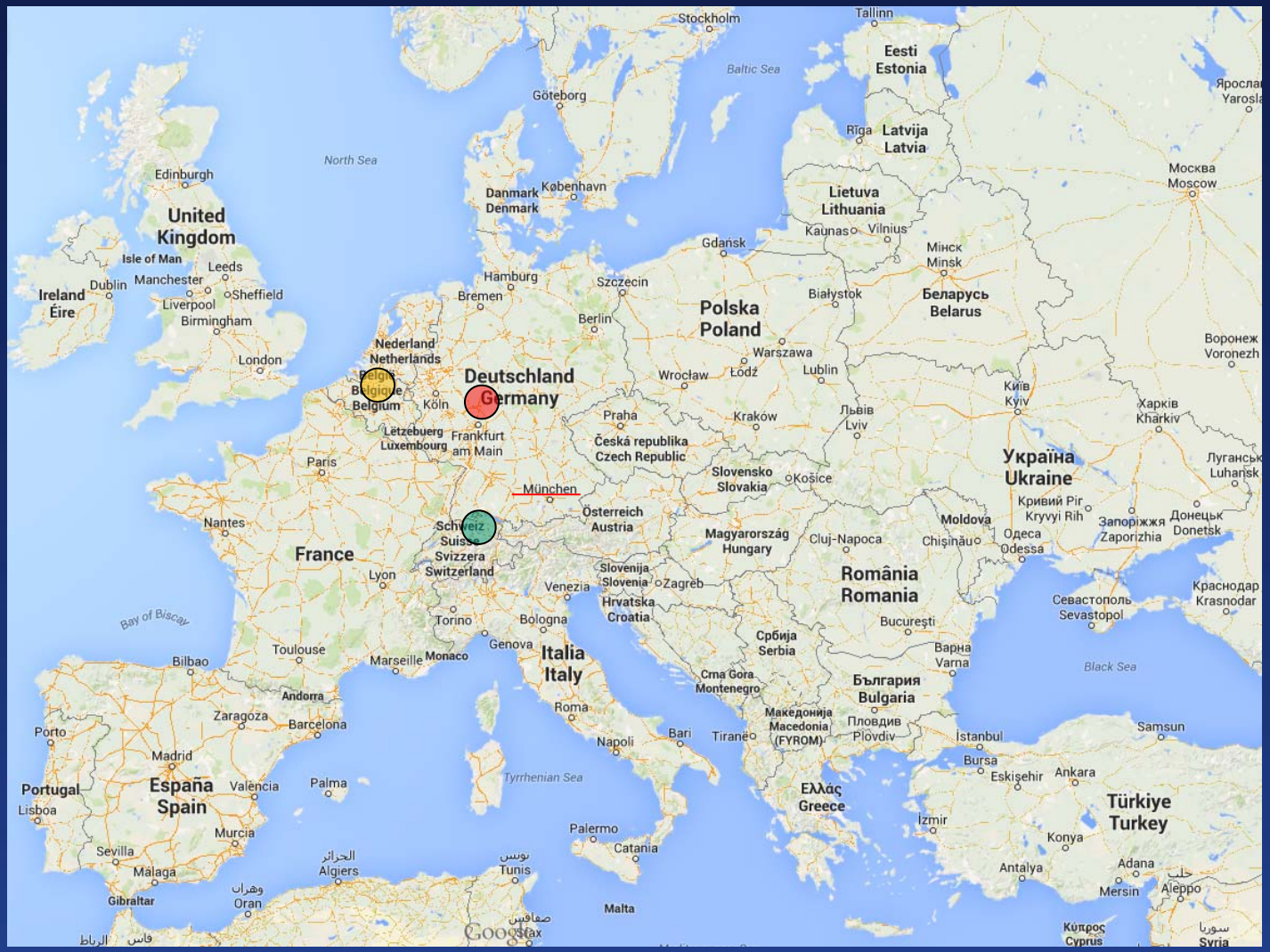
This is a serious accusation - especially for a german doctor.

MMC: Fetal surgery



- „DZFT“ Marburg Th. Kohl
- Leuven
- Zürich

Pränatal-Medizin München