Prenatal Diagnosis and Counseling on Myelomeningocele in Croatia

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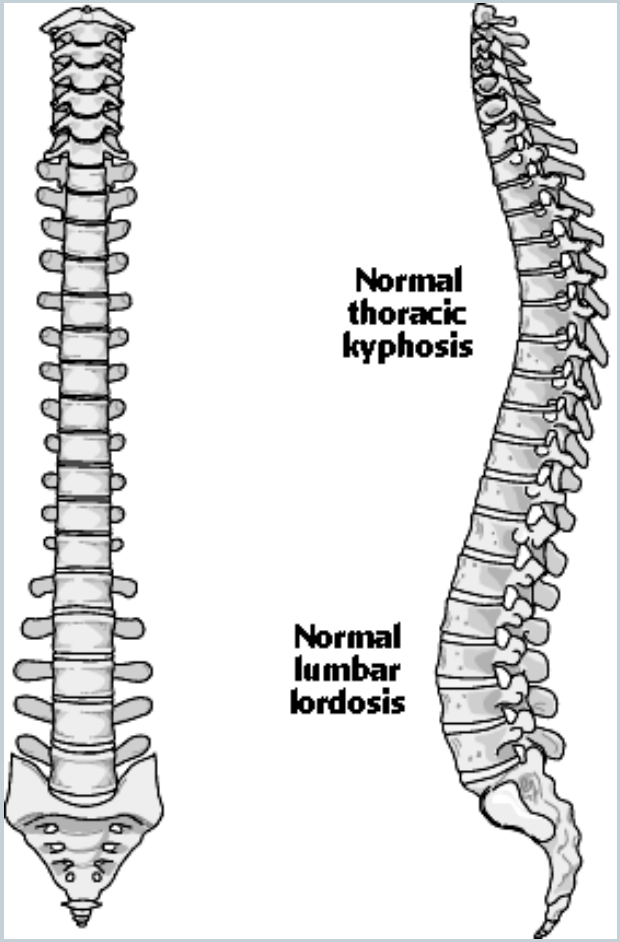
Normal spine

- Visualize (and document):
- US and MRI
  - Cervical, thoracic, abdominal, lumbar and sacral part
  - Transverse and longitudinal view
  - Triangular shape of osification centres (closed triangle)



MRI

- increasingly used in clinical practice, partly because of the increasing interest in fetal surgery and fetal medicine

- Ultra-fast MRI

Glenn OA and Coakley FV. MRI of the fetal central nervous system and body. Clin Perinatl 2009;36:273-300

Ultrasound examination (level 1)

- 12-14 weeks

- 18-22 weeks

Longitudinal

Transverse

Osification centres

Spina bifida – type of myelomeningocele

- Neural tube defect
  - Forms 5 - 6 weeks
  - Lumbar and sacrolumbar the most frequent
  - Incidence 0.5-2:1000 (Croatia 0.5/1000 livebirth)
  - Related to chromosomal anomalies
- Prognosis dependant on extent of the lesion
  - Possible total paralysis below the lesion
- Related to:
  - Hidrocephalus
  - Congenitalnital defecs
  - Meckel's syndrom (polidactily, multicystic kidneys - autosomal recessive)
  - Amnotic band sindrome (not genetic)

Spina bifida

- Classification

Spina bifida

- Closed
  - Covered by skin
  - Normal AFP

Spina bifida

- Open
  - Not covered by skin, drainage of the cerebrospinal liquor
  - Raised AFP
  - Rachishisis: complete open spine with herniation and/or destruction of neural spine

Spina bifida

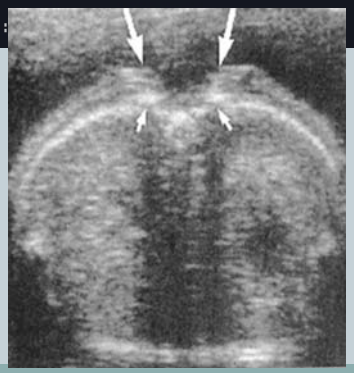
- Cystic
  - Meningocoele or meningomyelocoele (myelomeningocoele)

Spina bifida

- Ocult – not type of myelomeningocoele
  - Part of the vertebrae is not closed but without open defect
  - No clinical significance and without raised AFP

Dijagnosis

- US
  - Transversal plane the best for diagnosis but sagital better to assess the extent of the lesion
  - Protrusion of the sac (meninges) with spinal cord (or not)
  - Indirect signs (banana and/or lemon) – obliterated cisterna magna

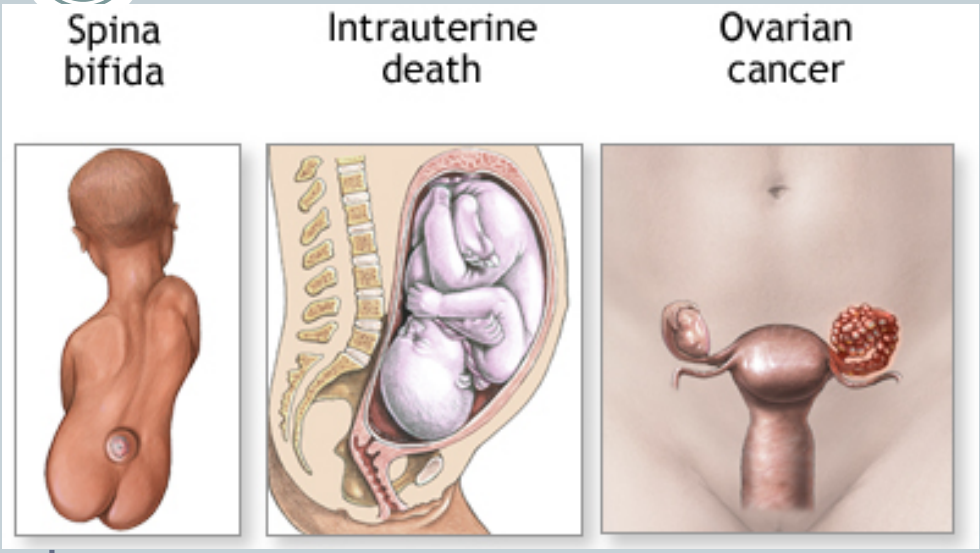


Diagnosis

- MRI
  - Since 2000
  - Rarely used in Croatia
  - Helpful..., problems in interperetation of immages
  - Compared to US

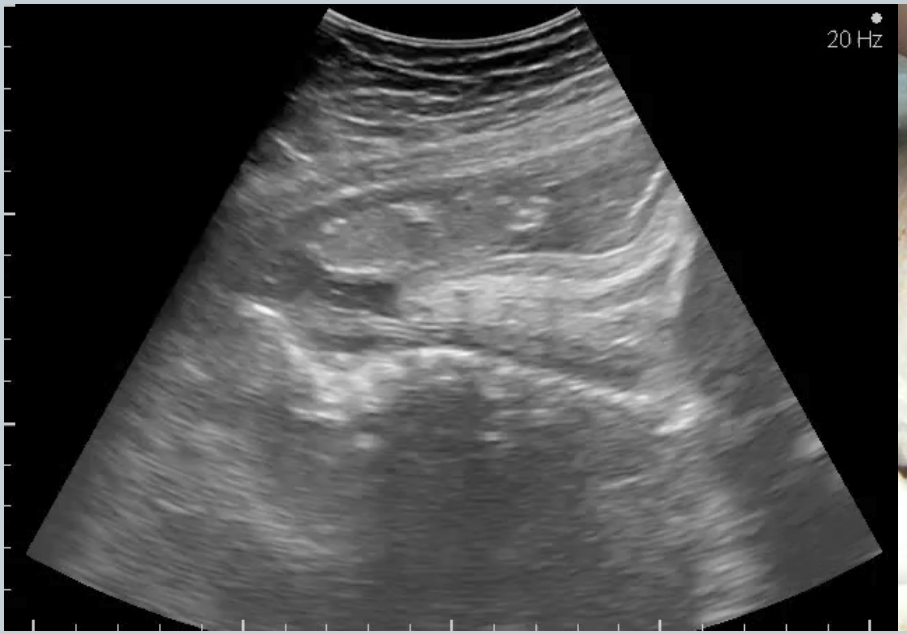
Dijagnosis

- AFP

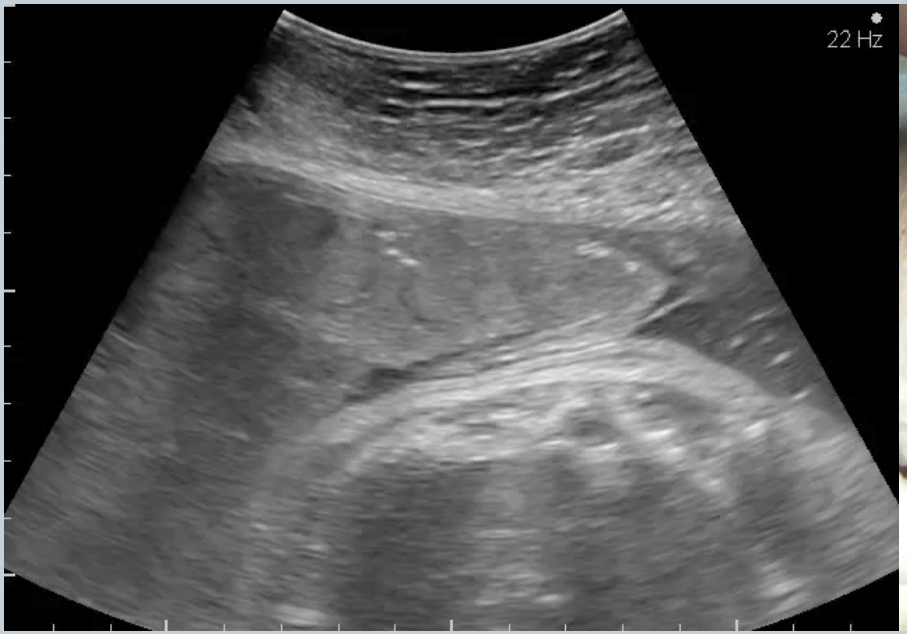


  - Not used any more in Croatia
  - Sensitivity 65%, combined with US gestational age assessment
  - US sensitivity up to 100% (P< .001 compared with screening) but later in pregancy
    - Dashe J i sur. AJOG 2006;195:1623

Spina bifida - transverse

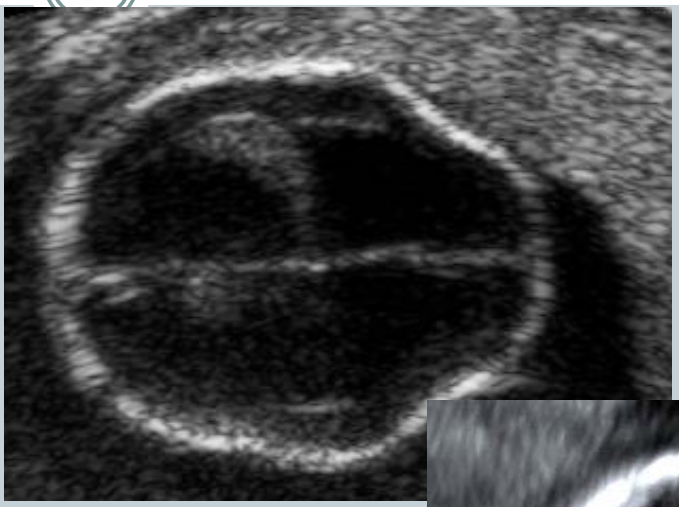


Spina bifida - sagital



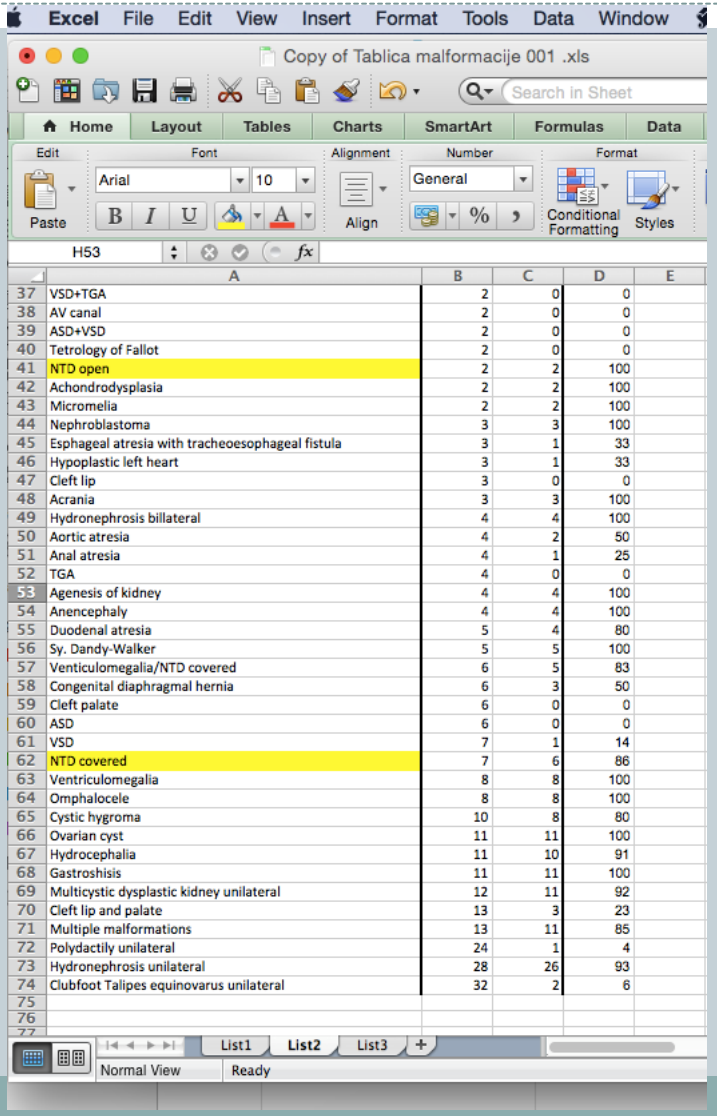
Associated anomalies

- Hidrocephalus, encephalocolela-e, Arnold Chiari
- Meningomielocoele (more frequent that meningocoele)
- Any level on fetal spine
- Abnormal osification of posterior centres



Ultrasound diagnosis - Croatia

- Incidence 0.5/1000 (livebirth) lower rate
- Diagnostic accuracy 50-100% (depending on centre)
- TOP if before 22/40
- Folic acid uptake different
- Referal centre



	A	B	C	D	E
37	VSD+TGA	2	0	0	
38	AV canal	2	0	0	
39	ASD+VSD	2	0	0	
40	Tetrology of Fallot	2	0	0	
41	NTD open	2	2	100	
42	Achondrodysplasia	2	2	100	
43	Micromelia	2	2	100	
44	Nephroblastoma	3	3	100	
45	Esphageal atresia with tracheoesophageal fistula	3	1	33	
46	Hypoplastic left heart	3	1	33	
47	Cleft lip	3	0	0	
48	Acrania	3	3	100	
49	Hydronephrosis billateral	4	4	100	
50	Aortic atresia	4	2	50	
51	Anal atresia	4	1	25	
52	TGA	4	0	0	
53	Agenesis of kidney	4	4	100	
54	Anencephaly	4	4	100	
55	Duodenal atresia	5	4	80	
56	Sy. Dandy-Walker	5	5	100	
57	Venticulomegalia/NTD covered	6	5	83	
58	Congenital diaphragmal hernia	6	3	50	
59	Cleft palate	6	0	0	
60	ASD	6	0	0	
61	VSD	7	1	14	
62	NTD covered	7	6	86	
63	Ventriculomegalia	8	8	100	
64	Omphalocele	8	8	100	
65	Cystic hygroma	10	8	80	
66	Ovarian cyst	11	11	100	
67	Hydrocephalia	11	10	91	
68	Gastroshisis	11	11	100	
69	Multicystic dysplastic kidney unilateral	12	11	92	
70	Cleft lip and palate	13	3	23	
71	Multiple malformations	13	11	85	
72	Polydactily unilateral	24	1	4	
73	Hydronephrosis unilateral	28	26	93	
74	Clubfoot Talipes equinovarus unilateral	32	2	6	
75					
76					

Spina bifida

- Folic?
  - Vit B
  - Reduces the risk up to 70% - BEFORE PREGNANCY
- Natural sources
  - Vegetables, fruit, juices
  - Insufficient
- How much?
  - 400 mcg (0.4 mg)
  - High risk (history) 4000 mcg (4.0 mg) three months

Ireland example

- 225 998 births, 236 NTD, 1.04/1 000
- 0.92/1 000 in 2009 to 1.17/1 000 in 2011
  - 45% (n = 106) had anencephaly
  - 49% (n = 115) had spina bifida
  - 6% (n = 15) had an encephalocoele
- Peri-conceptional folic acid supplement intake was 13.7% among the 52.5% (n = 124) of cases whose folic acid supplement intake was known.
- ?mandatory folic acid food fortification

McDonnel R et al. Neural tube dfects in the Republic of Ireland 2009-2011.J Public Health 2014

Something new?

- Neural tube defects (NTDs) have **not** significantly decreased in prevalence over the last 10 years, showing that efforts to prevent NTD with periconceptional folic acid supplementation have been largely unsuccessful.

Newsletter, 22nd december 2014

Spina bifida

- Antiepileptics
  - Valporate, filate use, increasing the dose
- Jentink et al. Does folic acid use decrease the risk for spina bifida after in utero exposure to valproic acid? Pharmacoepidemiolog Drug Saf 2010;19:803
  - OR – with folic acid

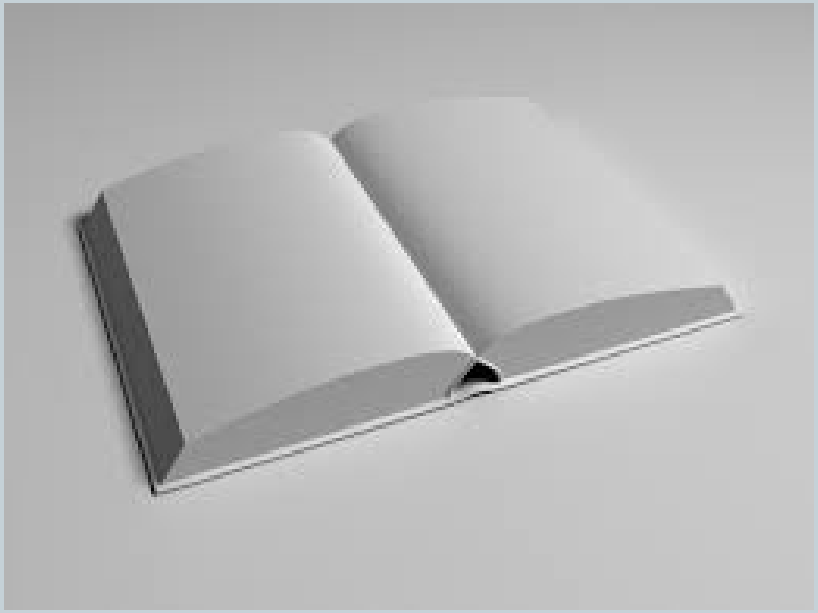
    not exponated 0.5 [95%CI: 0.3-0.7]

    eksponated 1.0 [95%CI: 0.1-7.6].
- Conclusion not possible
- Folic acid may not reduce the risk of spina bifida realted to valoprate use, but reduces the risk in general

- Inconclusive regarding the dose
- Croatia: “Why not?” principle, 10 times the normal dose

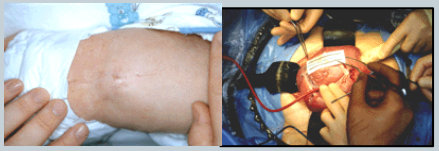
Spina bifida

Intrauterine surgery



www.nih.gov 2014

Adzik NS. Sem Pediat Sur, 2013



Spina bifida

LABOR AND DELIVERY

- CAESAREAN SECTION IS THE COMMONEST WAY, mandatory if breech, possible if there is no makrocephaly

Wilson RD et al J Obstett Gynaecol Can 2014;36:927

- Is Cesarean helpful regarding neonatal outcome – remains open question

Hill AE and Beattie F. Eur J Paediatr Surg 1994;4:32

- No benefit of Cesarean in there is no other indication

McLone DG and Bowman RM. Up-to-Date 2014

Croatia: Common sense... (menaing Caesarean)

Post natal manegement Croatia

- Postdelivery care - O & G
- Postnatal surgery – 5 centres
- On average 20 year (+/-3) HALFED IN RECENT YEARS
- Postnatal transfer

  Graz, Austria

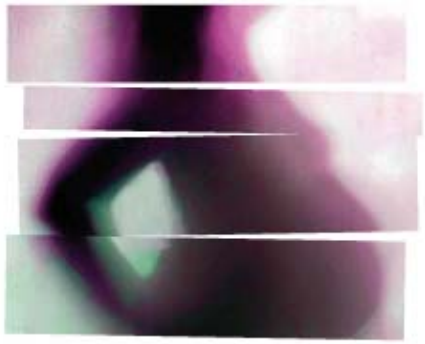
  (rare)

Colaboration

SVEUČILIŠTE U ZAGREBU MEDICINSKI FAKULTET

University of Zurich[UZH]

Thank you



6th Congress of the
South-East European
Society of Perinatal Medicine

Zagreb / CROATIA 4-6. 12. 2015.

The South East European Society
of Perinatal Medicine