NÖRAL TÜP DEFEKTLERİ

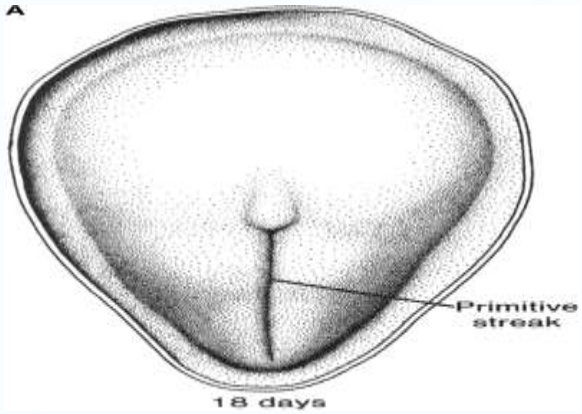
DR. GÖKHAN YILDIRIM

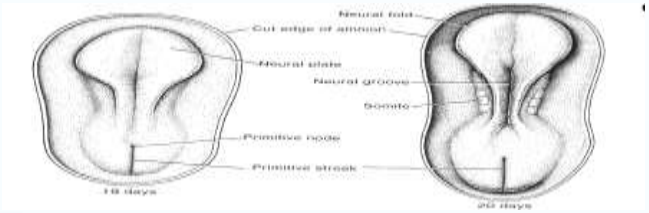
- En sık 2. konjenital anomali
- İnsidansı 1/1000 doğum
- Türkiye de insidansı ?
- Folik asit kullanımı insidansı azaltmıştır

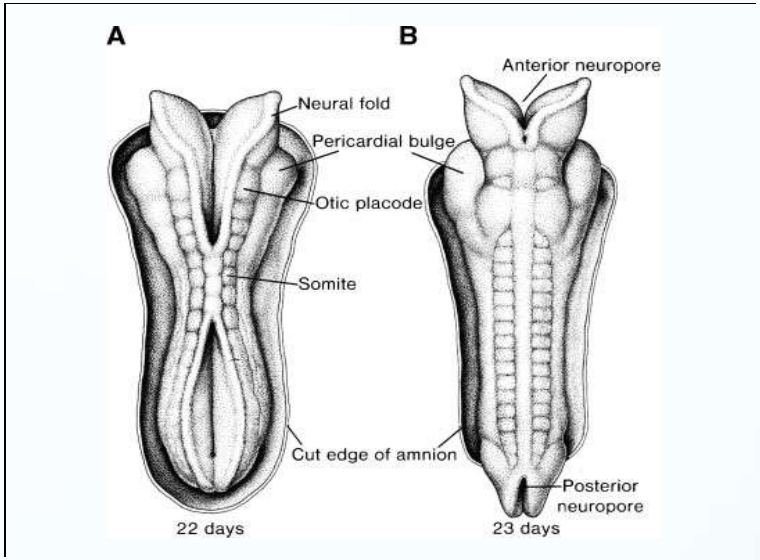
	Group 1 (2002-2006)	Group 2 (2007-2010)	Total	p
Central nervous system, n (%)	270 (59)	228 (45.2)	498 (51.8)	0.06
Multiple anomalies, n (%)	44 (9.6)	54 (10.7)	98 (10.2)	0.3
Chromosomal anomalies, n (%)	26 (5.7)	64 (12.7)	90 (9.4)	<0.0001
Genitourinary system n (%)	40 (8.7)	43 (8.5)	83 (8.6)	0.7
Musculosekeletal system, n (%)	30 (6.6)	39 (7.7)	69 (7.2)	0.3
Cardiovascular system, n (%)	8 (1.7)	26 (5.2)	34 (3.5)	0.002
Pulmonary system, n (%)	3 (0.7)	10 (2)	13 (1.4)	0.052
Gastrointestinal system, n (%)	7 (1.5)	6 (1.2)	13 (1.4)	0.8
Genetic diseases, n (%)	11 (2.4)	4 (0.8)	15 (1.6)	0.07
Face and neck, n (%)	7 (1.5)	7 (1.4)	14 (1.5)	1
Other, n (%)	12 (2.6)	23 (4.6)	35 (3.6)	0.06
Total, n (%)	458 (100)	504 (100)	962 (100)	-

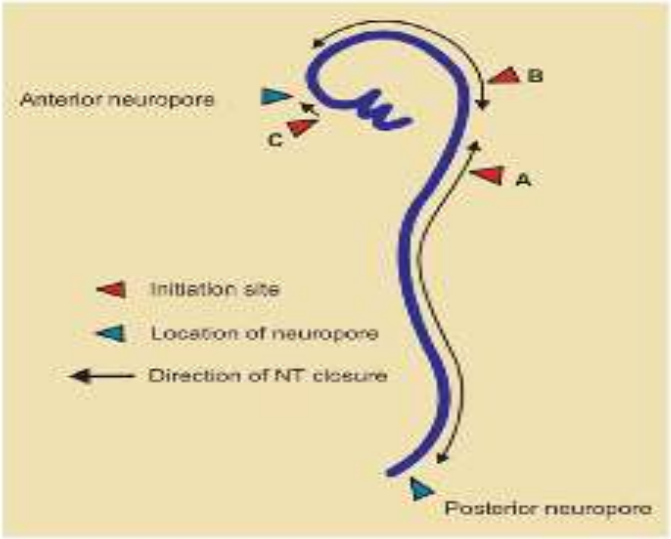
J Turkish-German Gynecol Assoc 2012; 13: 85-90

Embriyoloji



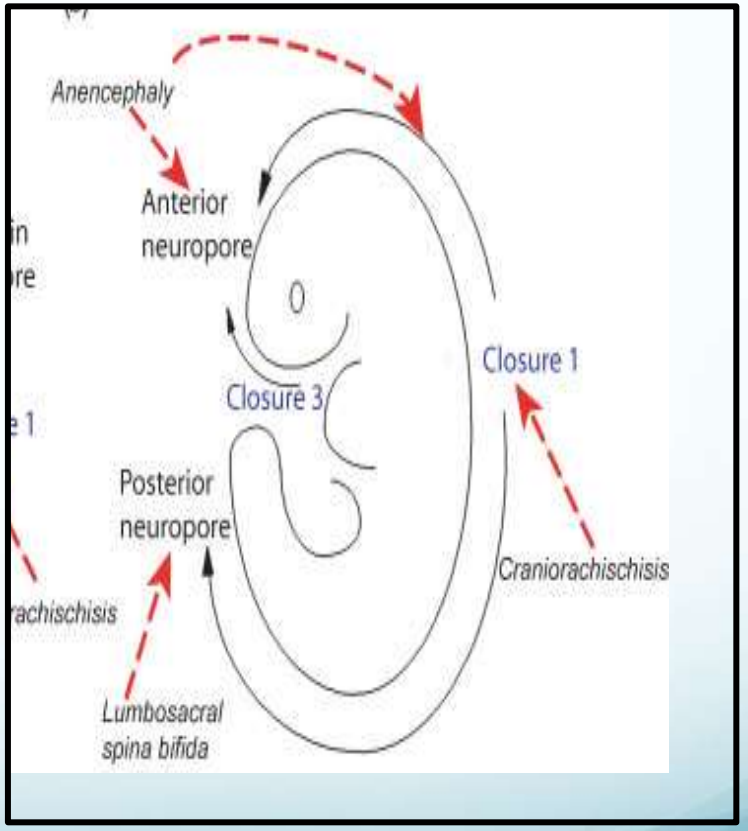






Sınıflama

- Kraniyal Defektler
  - Anensefali
  - Eksensefali
  - Ensefalosel
  - İniensefali
- Spinal defektler
  - Açık/kapalı
  - Meningosel/meningomiyelosel



Etiyoloji ve Risk Faktörleri

Genetik nedeler
- Aile öyküsü—multifaktöryel kalıtım
- MTHFR mutasyonu—677C→T
- Otozomal resesif kalıtımlı sendromlar
  - Meckel-Gruber
  - Roberts
  - Joubert
  - Jarcho-Levin
  - HARDE—hidrosefali-agri-retina displazisi-ensefalosel
- Anöploidi
  - Trizomi 13
  - Trizomi 18
  - Triloidi

Bazı çevresel ajanlara maruz kalmak
- Diabet—hiperglisemi
- Hipertermi
  - Jakuzi veya sauna
  - Ateş (tartışmalı)
- İlaçlar
  - Valproik asit
  - Karbamazepin
  - Kumadin
  - Aminopterin
  - Talidomid
  - Efavirenz

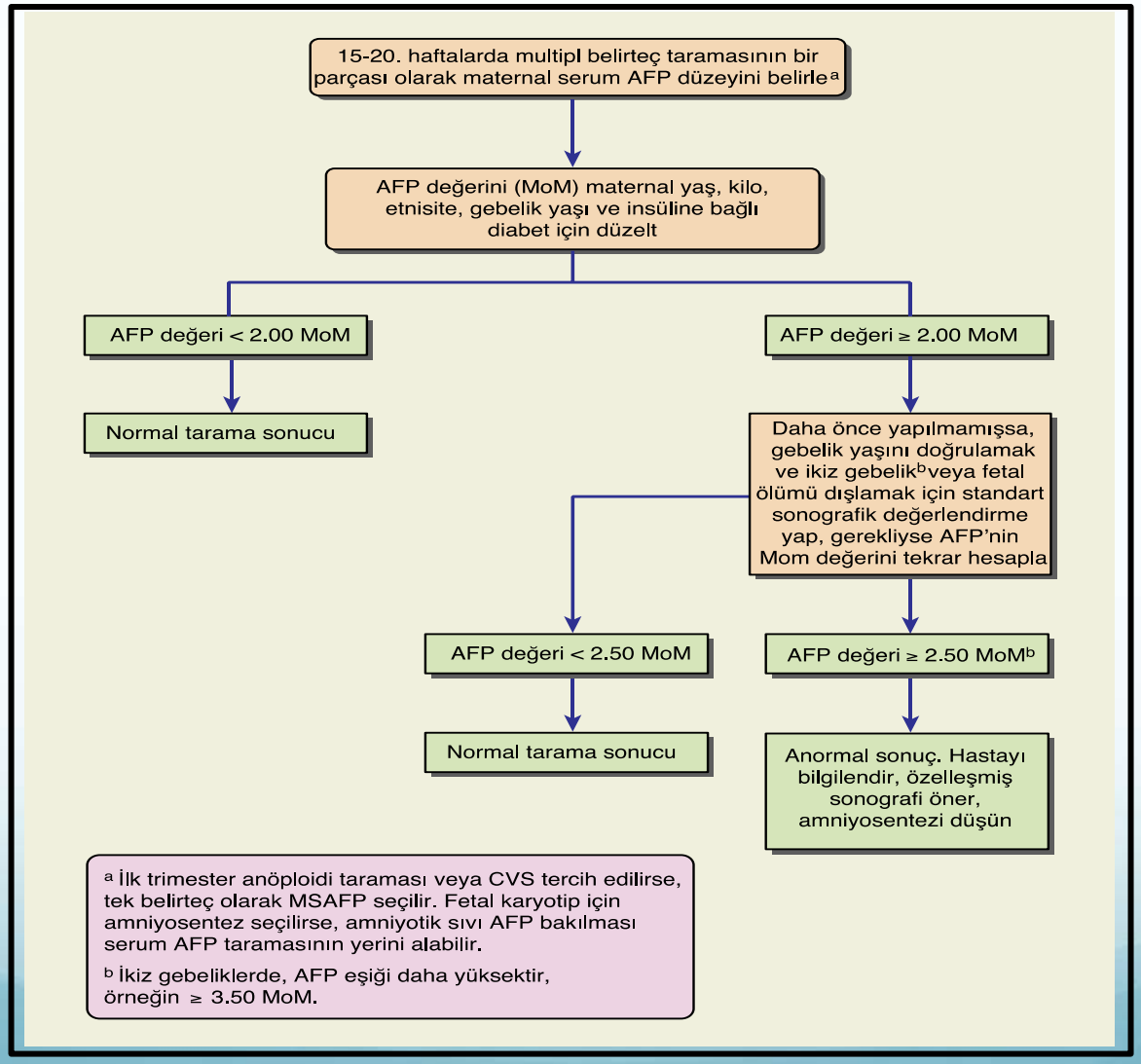
Coğrafi bölge—etnisite, diyet, diğer faktörler
- Birleşik Krallık
- Hindistan
- Çin
- Mısır
- Meksika
- Birleşik Devletlerde Güneydoğu Appalachian bölgesi

Tekrarlama Riski

Mother as reference	
General incidence	1.4-1.6
Women with diabetes mellitus	20
Women on valproic acid in first trimester	10-20
Fetus as reference	
1 sibling with neural tube defect	15-30
2 siblings with NTD*	57
Parent with NTD	11
Half sibling with NTD	8
First cousin (mother's sister's child)	10
Other first cousins	3
Sibling with severe scoliosis secondary to multiple vertebral defects	15-30
Sibling with occult spina dysraphism	15-30
Sibling with sacrococcygeal teratoma or hamartoma	~15-30

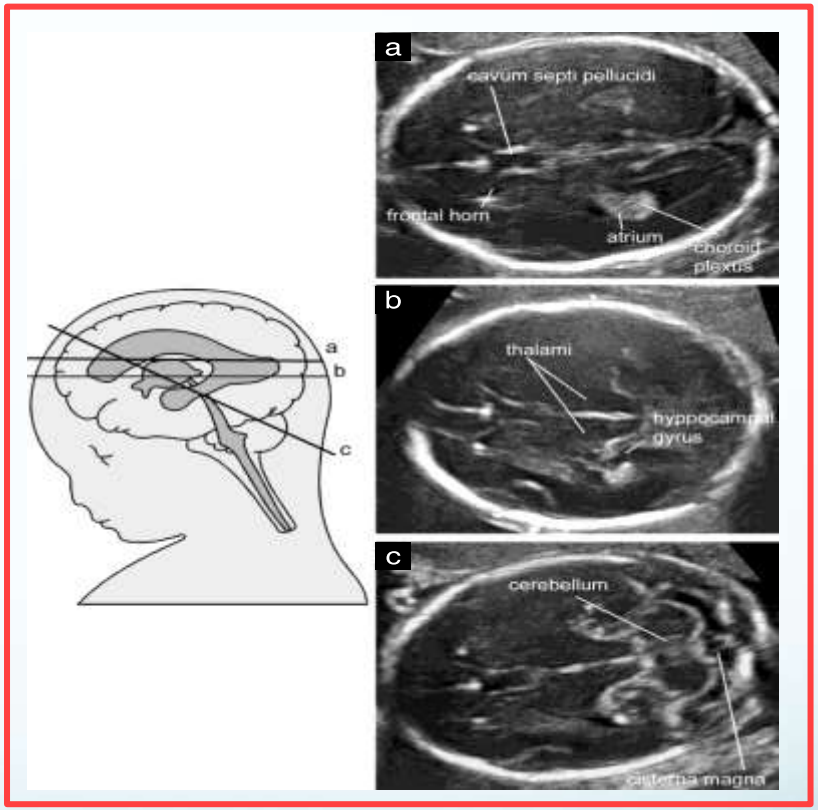
Tarama

Biyokimyasal Tarama



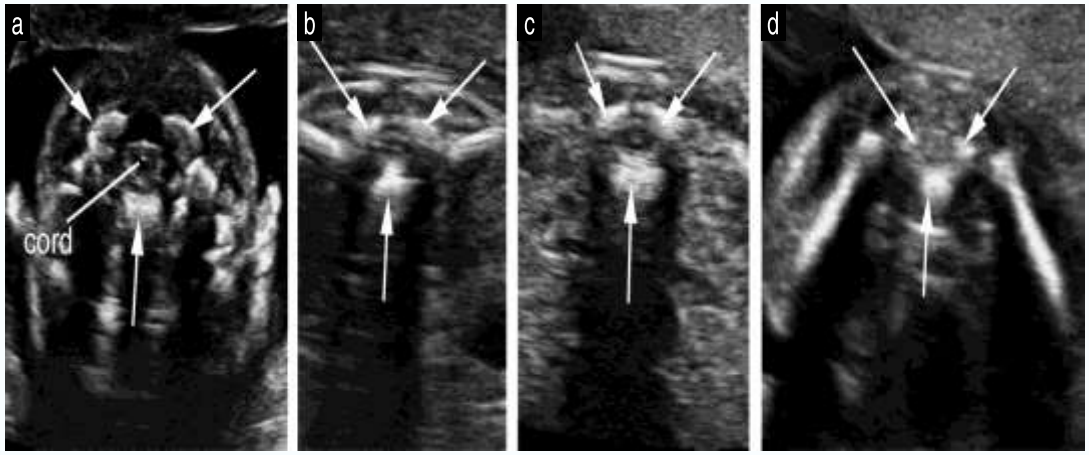
Ultrasonografi

Kraniyal Değerlendirme

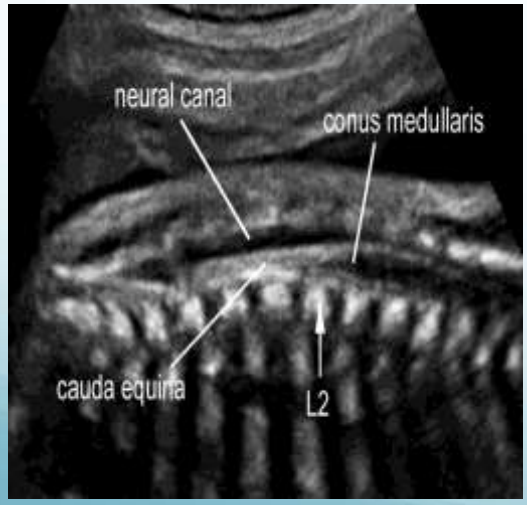


Spinal Değerlendirme

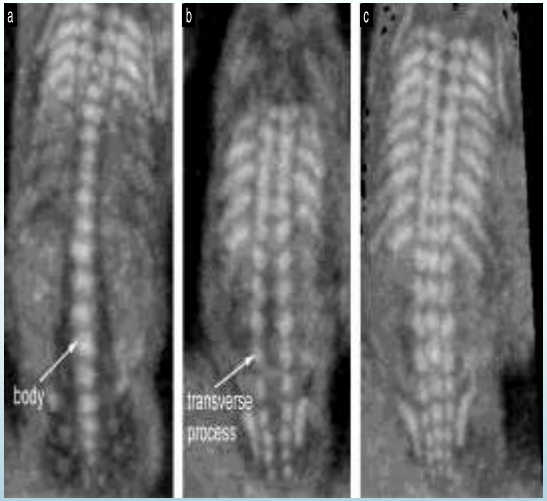
Transver/aksiyel kesit



Sagittal kesit



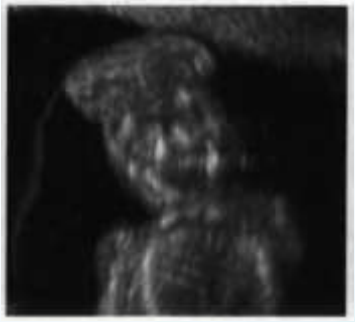
Koronal kesit



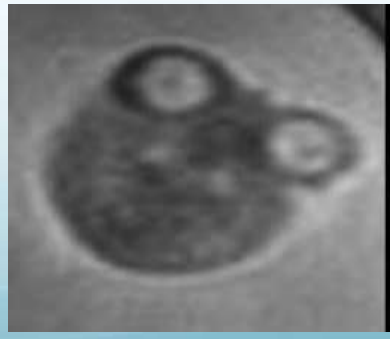
PRENATAL TANI

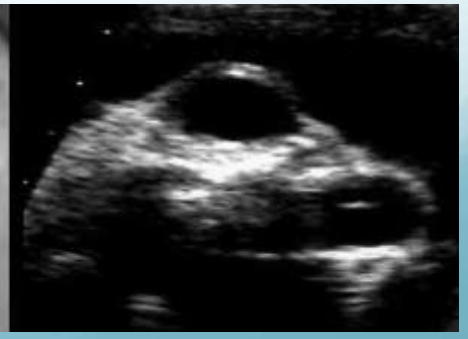
Eksensefali – Anensefali

- **İlk trimester USG bulguları:**
  - **Hemisferler (+)**
  - **Mickey Mouse bulgusu (+)**
  - **CRL küçük**
  - **Kranial kutup torakstan küçük ve düzensiz**
  - **Amniyon sıvısı ekojen**



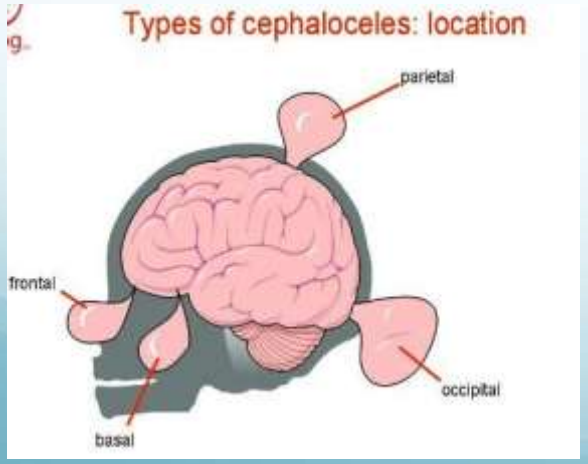
- İkinci trimester USG bulguları:
  - Kranyumun üst kısmı izlenmez
  - Heterojen kitle görünümü
  - Polihidroamnios

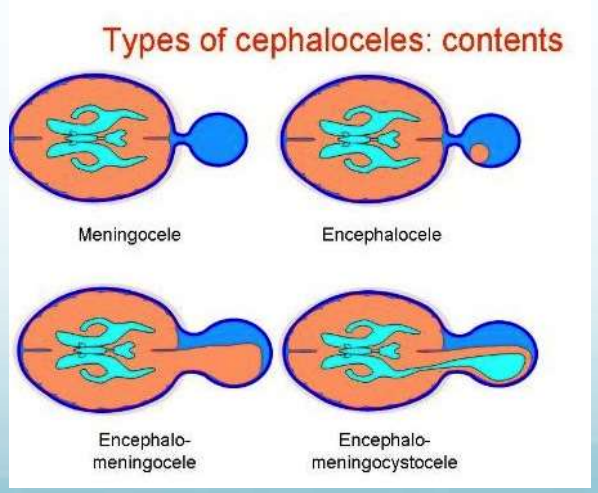




Ensefalosel/Sefalosel

- İntrakranyal içeriğin karanyal defektten protrüzyonu
- Sonografik bulgular;
  - Kistik ve/veya solid kitle
  - Kalvaryumda defekt
  - Kist içinde kist
  - Ventrikülomegali
- Karyotip





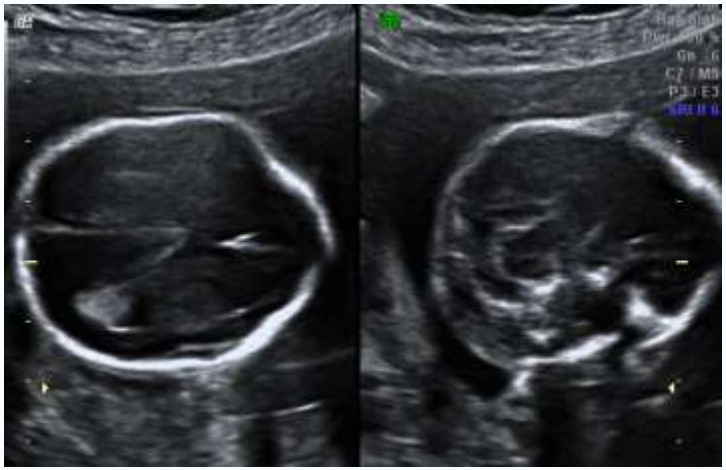
İniensefali

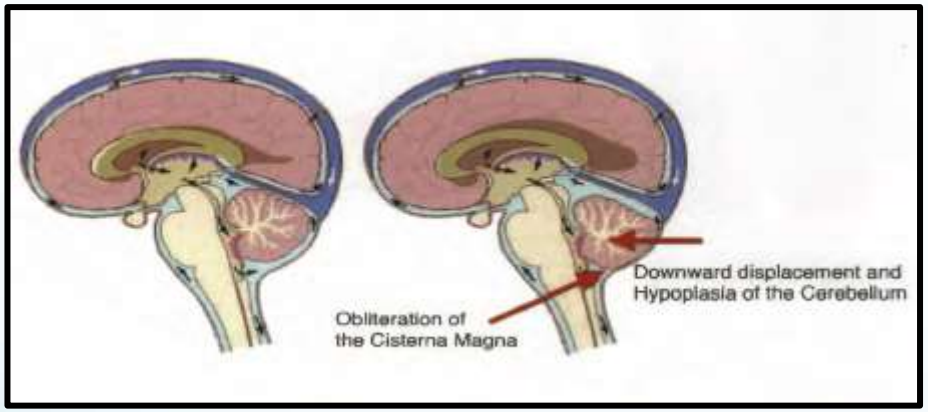
- Kompleks ve lethal bir malformasyon
- 3 temel özelliği var;
  - Foramen magnumu tutan oksiput defekt
  - Spinada retroflaksiyon
  - Açık spina bifida

Spina Bifida

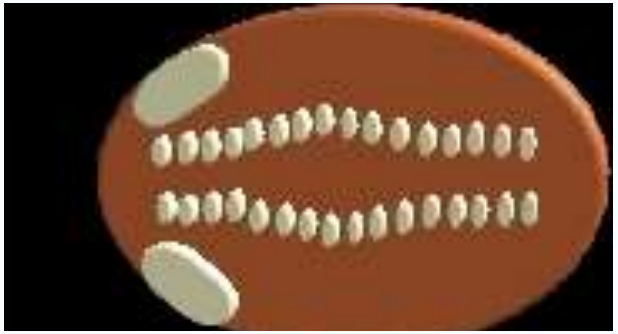
- Spinadaki kemik defekti
- Primer nörolasyon kusurlu
- İnsidansı 0.5-1/1000 gebelik
- Tipleri
  - Açık %85 – 90 (miyelosel/miyelomeningosel)
  - Kapalı %10 – 15
- %80'ni lumbar, torakolumbar veya lumbosakral
- Prenatal sonografinin sensitivitesi %97-98; spesifitesi %100

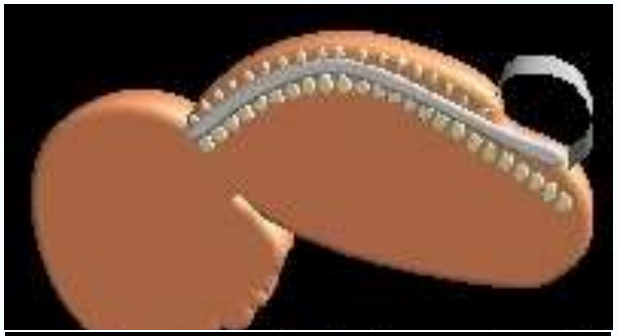
- **İntrakranyal bulgular;**
  - Mikrosefali
  - Limon belirtisi
  - Muz belirtisi
  - Ventrikülomegali
  - Chiari II malformasyonu





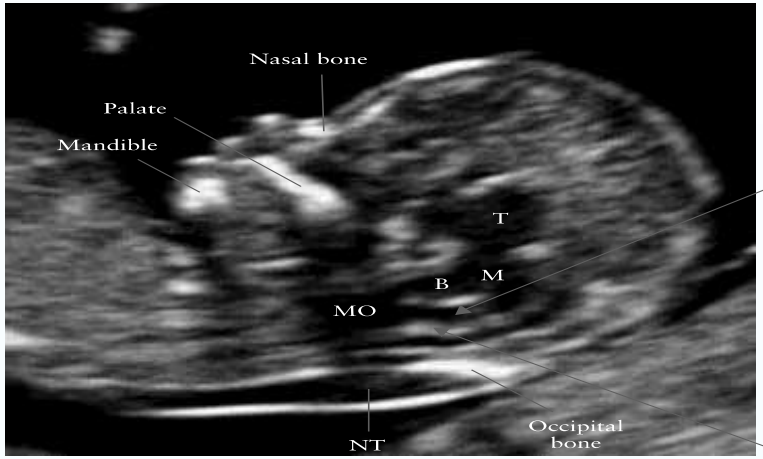
- **Spinal bulgular;**
  - Dorsal vertebral yapılarda genişleme
  - Meningosel kesesi
  - Miyelomeningosel kesesi
  - Kifoskolyoz

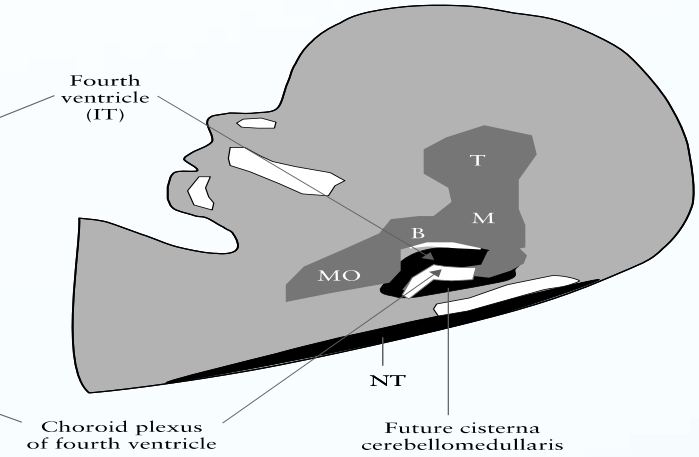


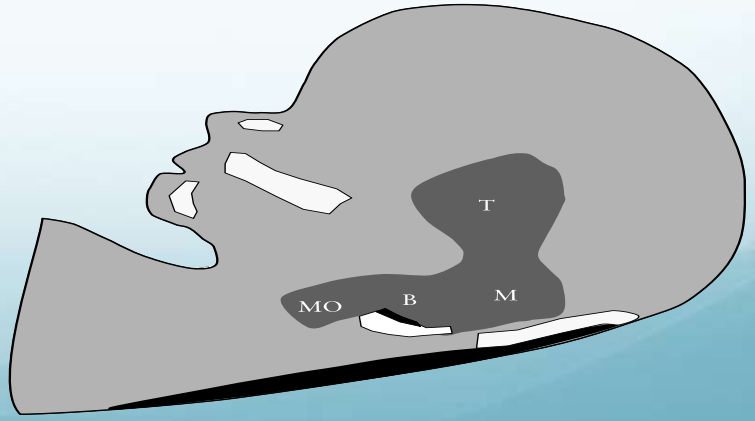


İlk Trimesterde Tanı

İntracranial Translucency







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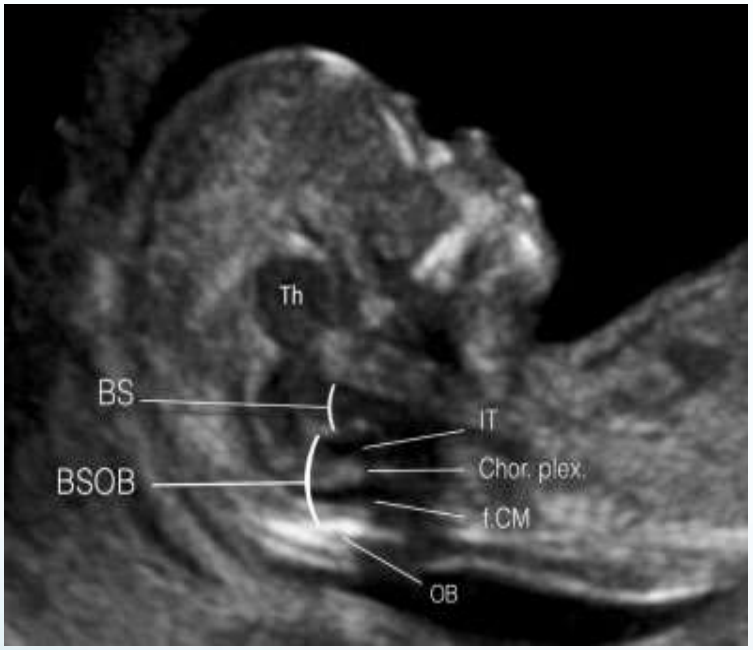
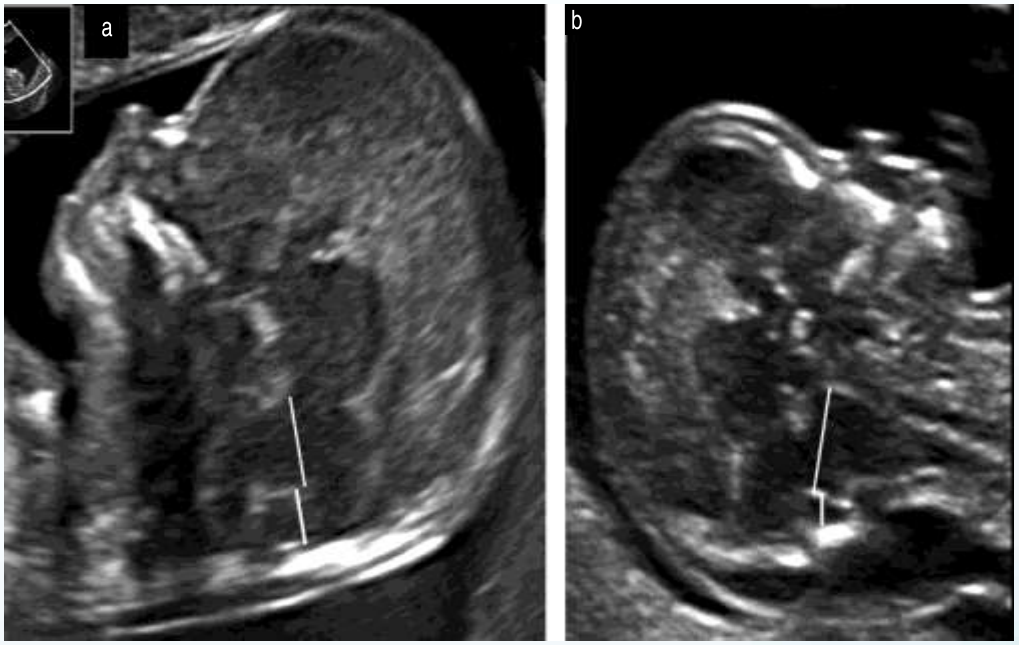
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