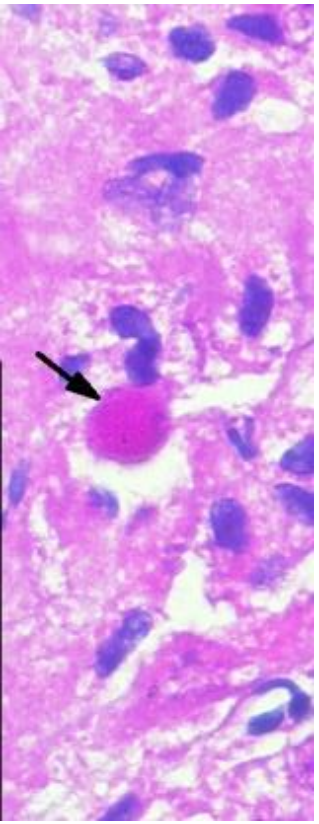
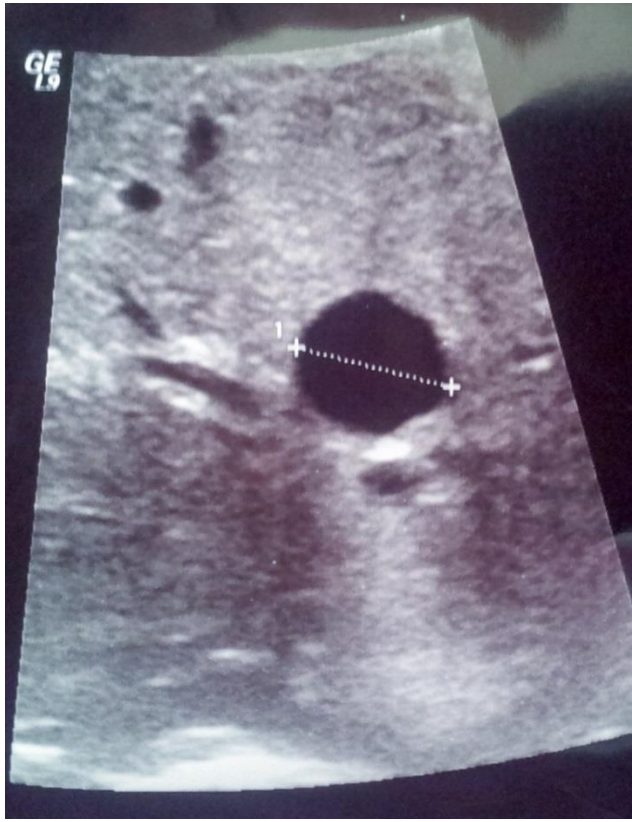
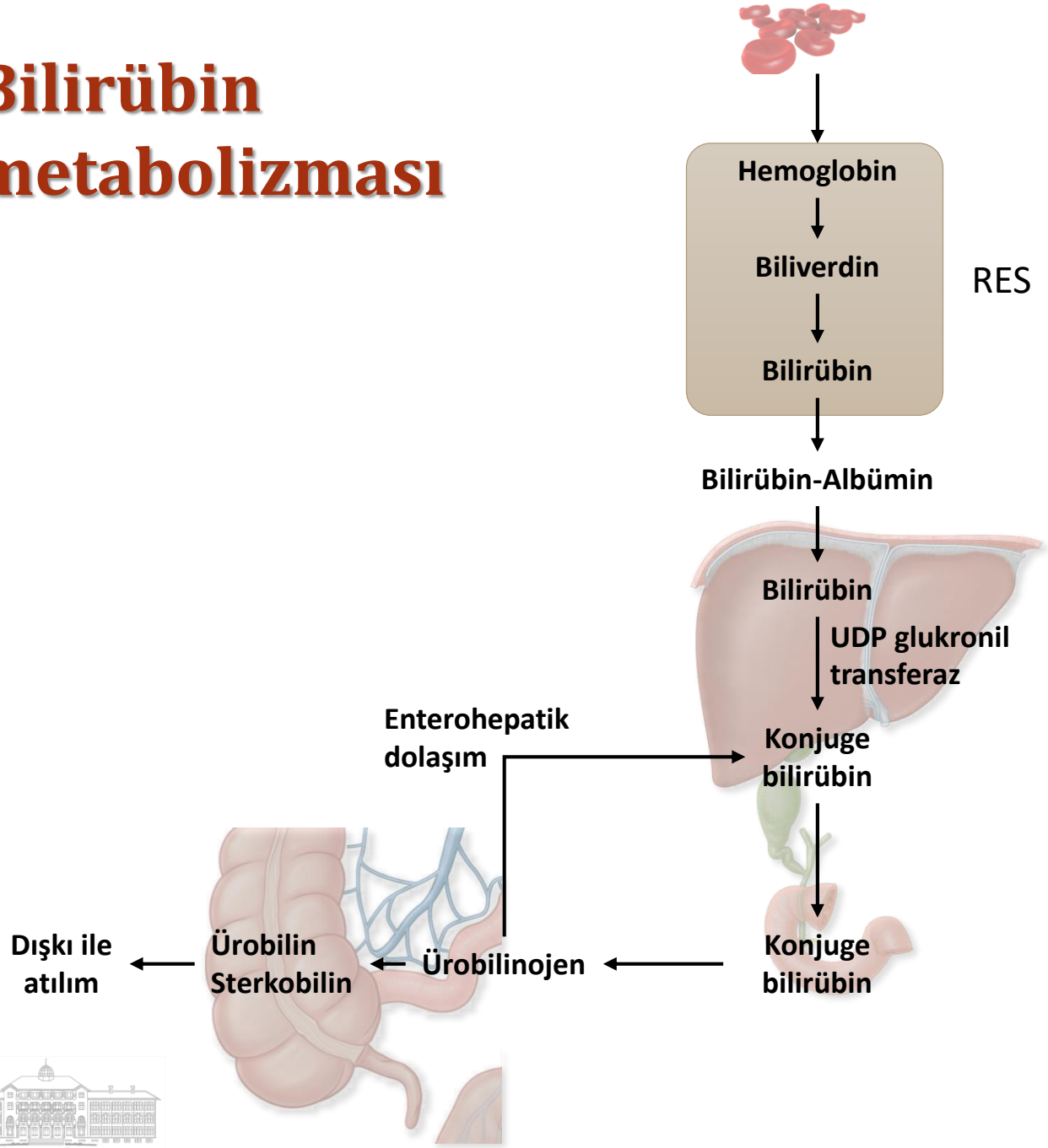


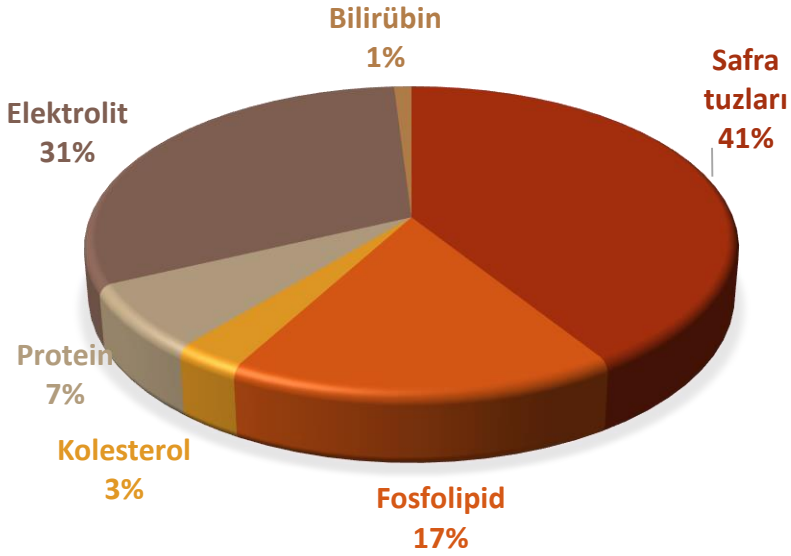
Direk bilirübinemisi olan bebek

Doç.Dr. Sinan Sarı
Gazi Üniversitesi Tıp Fakültesi
Çocuk Gastroenteroloji Bilim Dalı

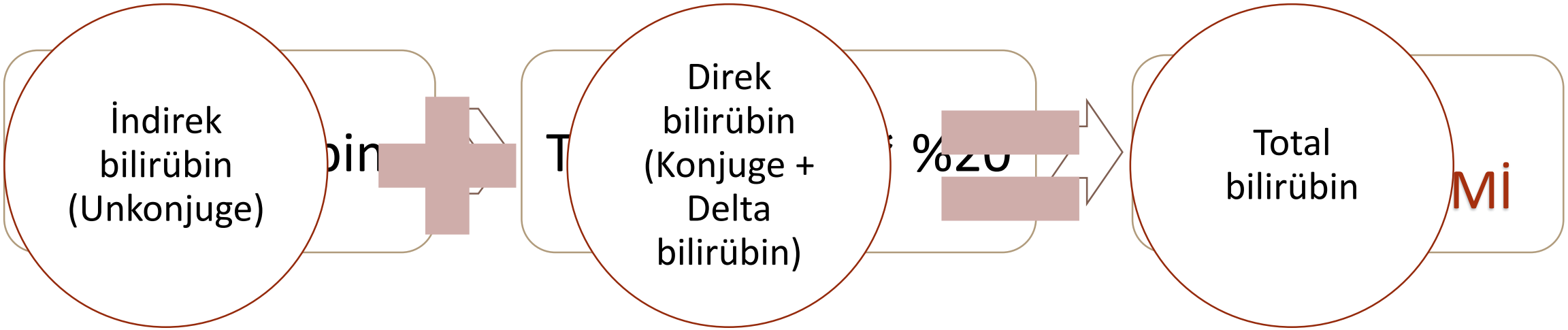


Bilirübin metabolizması





Direk bilirübinemi



Kolestazın biyokimyasal bulgusudur

JPGN 2004;39:115-128

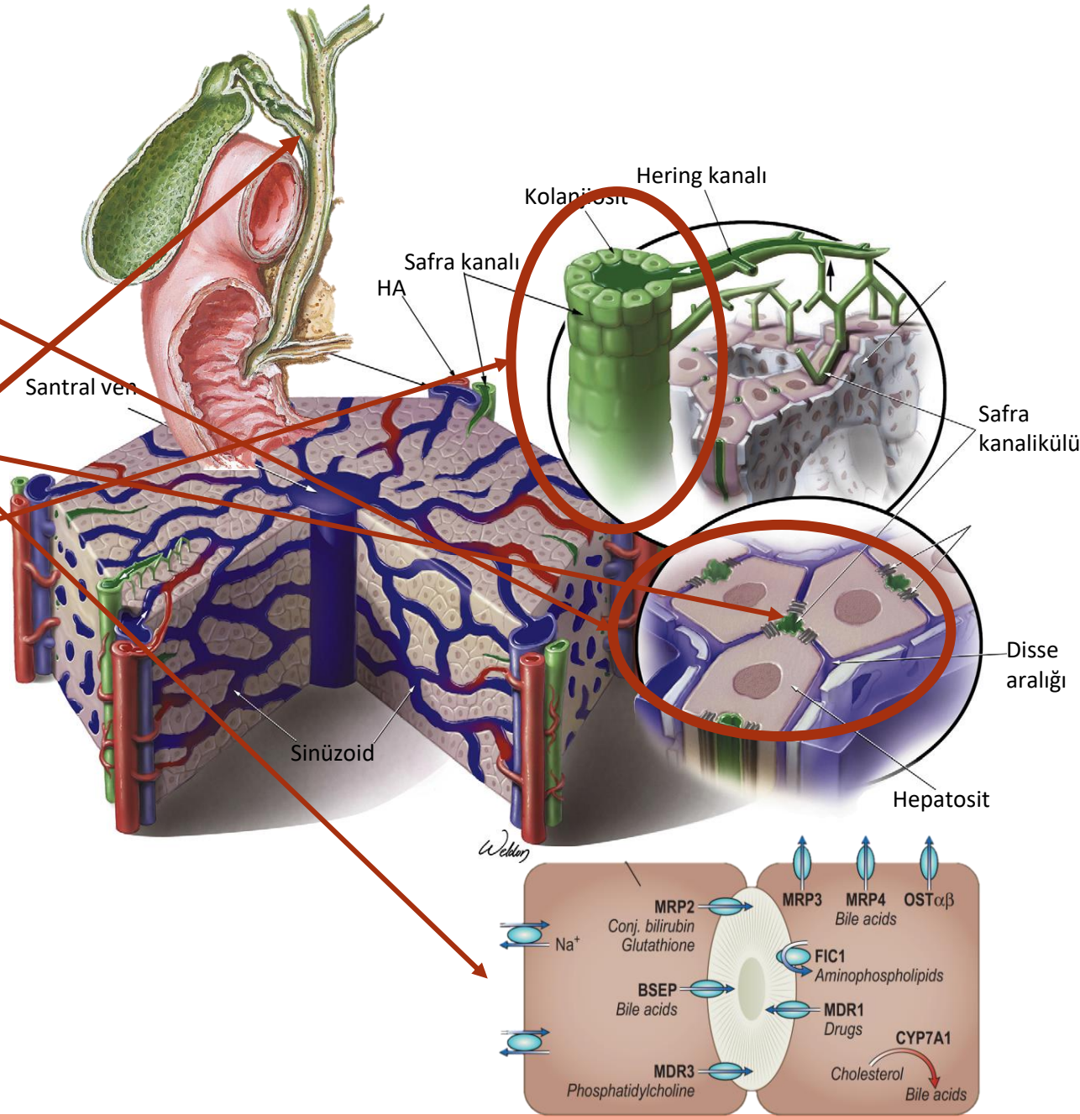
KOLESTAZ

Safra üretiminde azalma (hepatoselüler hasar)

Kanaliküler membranda transport sorunları

Safra akımında azalma

Safra ile atılan bilirübin, safra asitleri, kolesterol ve diğer maddelerin birikimi



KOLESTAZ

Retansiyon/
Regürjitasyon

- Safra asidi → **Kaşıntı** / Hepatotoksisite
- Kolesterol → Ksantomlar
- **Bilirübin** → **Sarılık**
- Bakır → Hepatotoksisite (?)

Progresif biliyer siroz

Son dönem karaciğer hastalığı
Portal hipertansiyon (asit vb.)

Bağırsağa safra salgısında azalma

Lümende azalmış safra asidi

Malabsorpsiyon → İshal

- Malnütrisyon
  Büyüme geriliği
- Yağda eriyen vitamin eksikliği
  - **A** → Gece körlüğü
  - **D** → Metabolik kemik hastalığı
  - **E** → Nöromuskuler dejenerasyon
  - **K** → Hipoprotrombinemi
- Kalsiyum eksikliği → Metabolik kemik hastalığı

Bebeklerde direk bilirübinemi nedenleri

- Biliyer hastalıklar
- Safra transport bozuklukları
- Enfeksiyonlar
- Metabolik hastalıklar
- Endokrinolojik hastalıklar
- İmmunolojik hastalıklar
- Genetik
- Hematolojik-Onkolojik
- Toksik
- Vasküler
- İdiyopatik neonatal hepatit

EKSTRAHEPATİK

- **Kistik fibrozis**
- **Alfa-1 antitripsin eksikliği**
- CH: **Galaktozemi**, Fruktozemi, tip IV glikojen depo
- LİPİD: Niemann-Pick tip C, Gaucher, Wolman
- AA: **Tirozinemi**, mevolanat kinaz eks.
- Üre siklus bozuklukları: **Sitrin eksikliği**
- Safra asit metabolizma bozuklukları
- Konjenital glikolizasyon defektleri
- Peroksizomal hastalıklar
- Mitokondrial hastalıklar

Direk bilirübinemi

- 1/2500
- Kolestazı erken tanımak
- Nedeni bulmak
- Başarılı tedavi
- İYİ PROGNOZ

- Geç danışım (en az %50)
- Yetersiz bebek izlemi
- Geç izlem
- Farkındalık az
- Bebekler sağlıklı, iyi kilo alıyor

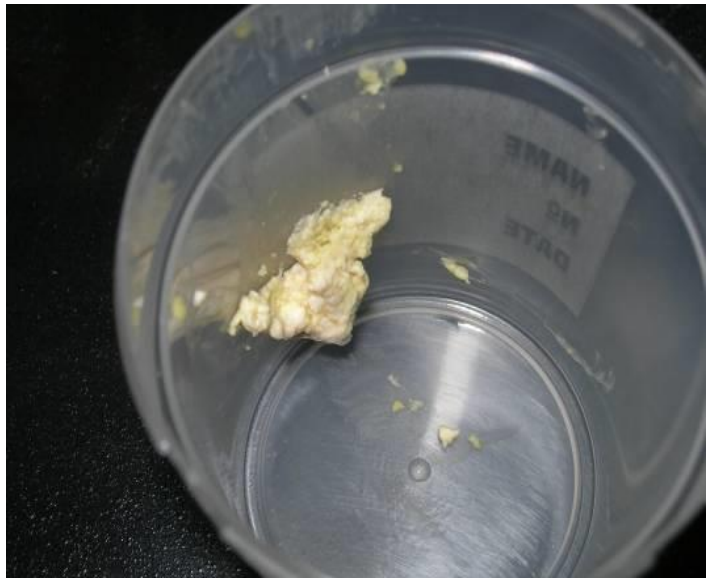
Uzamış sarılıklarda (>14 gün) mutlaka bilirübin fraksiyonları bakılmalı*
Erken tanı=Erken tedavi=Düşük komplikasyon

Öykü

- Sarılığın başlangıç zamanı ve süresi
- Akolik dışkı
- İdrarda koyulaşma
- Dışkı rengi kartları
- **Beslenme:** oral alımı; AS, mama, diyette fruktoz; emmeme, beslenme intoleransı; kusma; kilo alımı
- İshal
- Huzursuzluk
- Letarji
- Nöbet öyküsü
- Kanama
- Maternal hastalık, perinatal öykü
- Gebelik kolestazı
- Doğum öyküsü, doğum ağırlığı
- ABO, Rh uygunsuzluğu
- Pozitif aile öyküsü
- Akrabalık

Fizik inceleme

- Genel durumun değerlendirilmesi: letarji, huzursuzluk vb.
- Antropometrik ölçümler
- Nedene ilişkin bulgular
  - Mikrosefali
  - Dismorfik bulgular
  - Üfürüm, siyanoz
  - Katarakt: metabolik hastalık, konjenital enfeksiyonlar
  - Hepatomegali
  - Splenomegali
  - Peteşi, purpura, veziküler döküntüler (HSV)



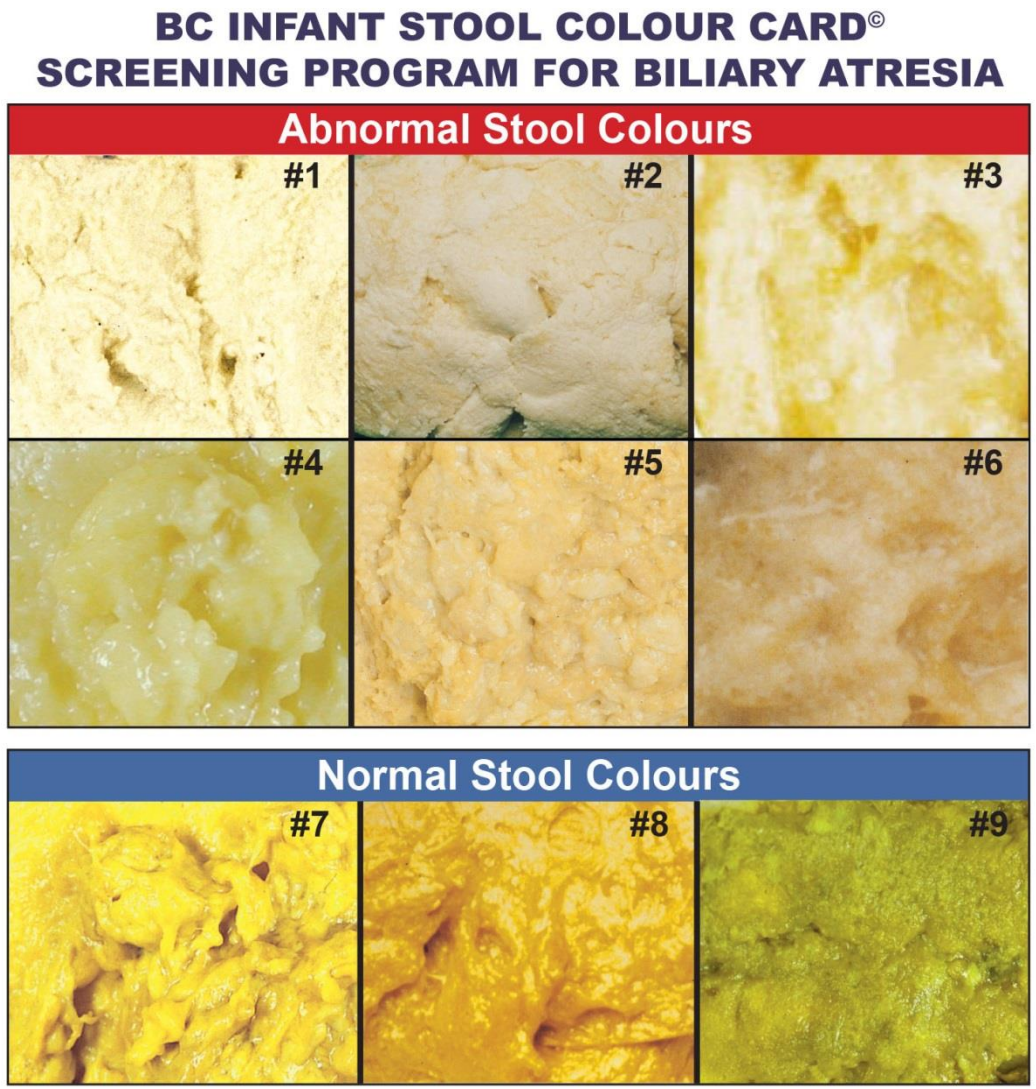
Fizik inceleme

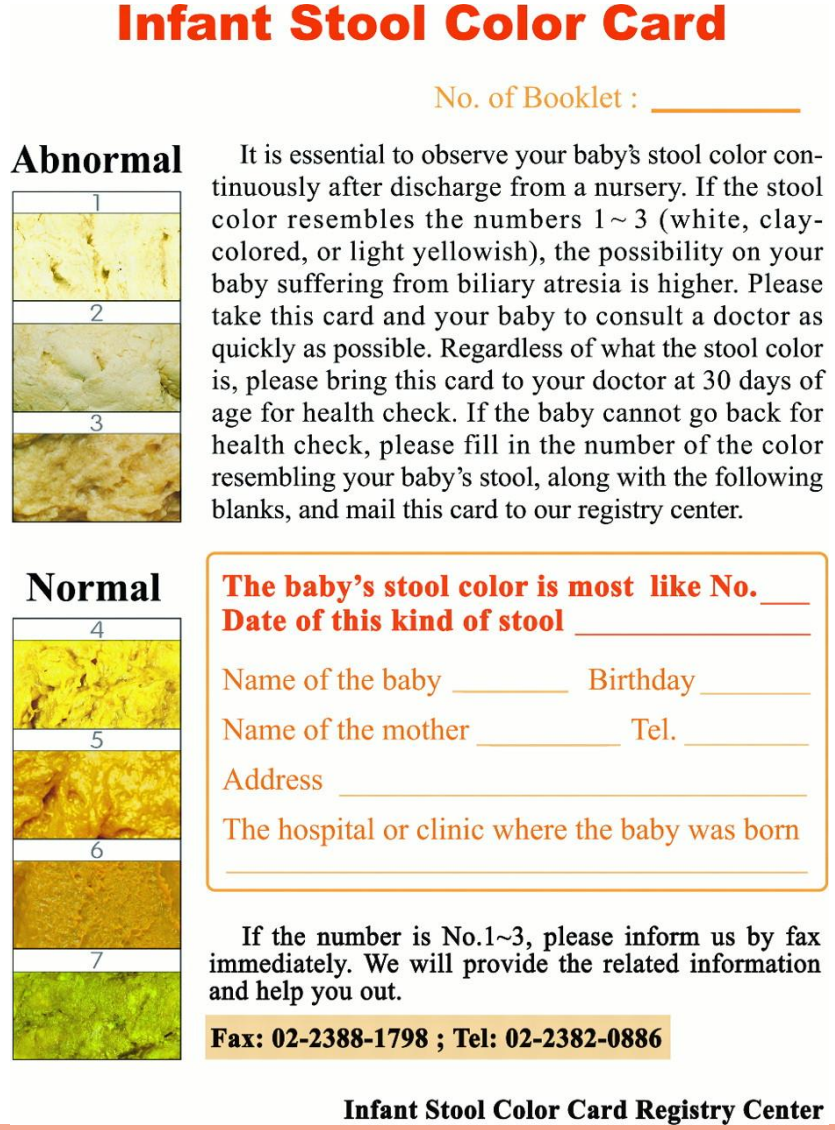
- Komplikasyonlara ait bulgular
  - Koagulopati
  - Portal hipertansiyon
  - Trombositopeni
  - Kilo alımında azlık
- Karaciğer yetmezliğinin bulguları
  - Ödem, asit
  - Koagulopati
  - Ensefalopati

Akut hasta bebek →

Enfeksiyonlar (TORCH, sepsis)
Metabolik hastalıklar
Hipopitüitarizm
Şok
Kalp yetmezliği

Dışkı kartları





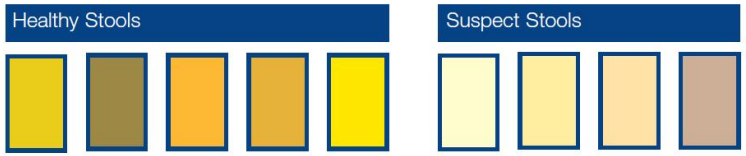
Yellow Alert

One of these babies has liver disease. Can you tell which one?

Difficult to decide? What can you do?

Check your baby's urine colour
If yellow — tell your doctor, health visitor or midwife

Check your baby's stool colour:



Stools suspect — tell your doctor, health visitor or midwife

If your baby is still jaundiced two weeks after birth (three in a premature baby) ask for a split bilirubin test

Need more information?
Contact Children's Liver Disease Foundation on
0121 212 3839
yellowalert.org

Scan with your smartphone and visit CLDF's Yellow Alert website

Join the conversation…
Follow @tweetcldf
Find us on Facebook – search CLDF

Children's Liver Disease Foundation
fighting childhood liver disease

36 Great Charles Street
Birmingham B3 3JY
Telephone: 0121 212 3839

childliverdisease.org
cldf-focus.org
info@childliverdisease.org
Registered Charity No. 1067331

Yellow Alert

Download on the App Store

ANDROID APP ON Google play

16:40

Children's Liver Disease Foundation

fighting childhood liver disease

16:41

Information

Jaundice can affect up to 90% of newborn babies and usually occurs two or three days after birth. In most cases jaundice will peak at around day 4 of life and then gradually disappear by the time the baby is two weeks old. Jaundice is NOT a liver disease and does not necessarily indicate that a baby is ill.

Jaundice that continues beyond 14 days of age in a full term baby or 21 days in a premature baby should be investigated immediately.

If a baby's stools and urine are not a healthy colour then this should be investigated at whatever age and shouldn't wait until the baby is two or three weeks old.

Prolonged Jaundice

16:42

Tests and Referrals

All babies with prolonged jaundice should have a split bilirubin test.

Any baby with jaundice persisting longer than 14 days from birth in full term babies or 21 days in pre-term babies and/or suspect colour stools or urine should be referred for a split bilirubin test straight away.

Levels of bilirubin and next steps

Level of conjugated bilirubin	Total bilirubin	Next steps
<20% (25 micromoles/l)	Less than 200 micromoles/l	Review weekly until levels return to normal
<20% (25 micromoles/	More than 200	Refer to paediatrician for follow up

16:41

Stool Chart

Persistently yellow urine staining the nappy can be a sign of liver disease.

Persistently pale coloured stools may indicate liver disease.

Click on any of the colours below to enlarge.

Healthy Stools

H1

H2

H3

H4

H5

16:42

Order a Pack

The Yellow Alert App is supplementary to the Yellow Alert Pack. CLDF's Yellow Alert Pack has been put together to help in the identification and referral of prolonged jaundice in newborn babies.

The Yellow Alert Pack contains:-

- **Jaundice Protocol** - A protocol written for healthcare professionals to provide general

Laboratuvar

- Enzimler
  - **AST**
  - **ALT**
  - **GGT**
  - **ALP**
- Sentez fonksiyonları
  - **PT**
  - **Albümin**
  - **Glukoz**
  - Amonyak

- AST, ALT
  - Kalp ve iskelet kası, böbrek, pankreas, eritrositler
  - Koenzim: B6
- ALP
  - Kemik, böbrek, ince barsak
  - Koenzim: Zn
- GGT
  - Pankreas, dalak, beyin, meme, ince bağırsak, böbrek
- ALP, GGT
  - **YAŞA ÖZEL NORMAL DEĞERLER ÖNEMLİDİR**

GGT normal kolestazlar

PFIK 1 ve 2
Safra asit metabolizma boz.
Artrogripozis
MVID
Hipopitüitarizm

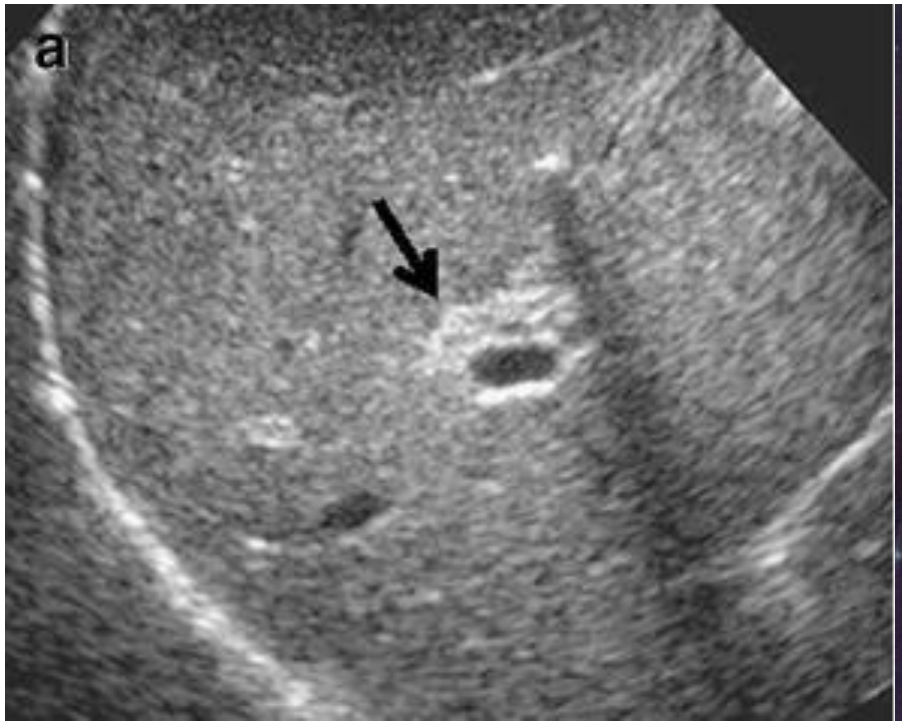
DİĞER TESTLER

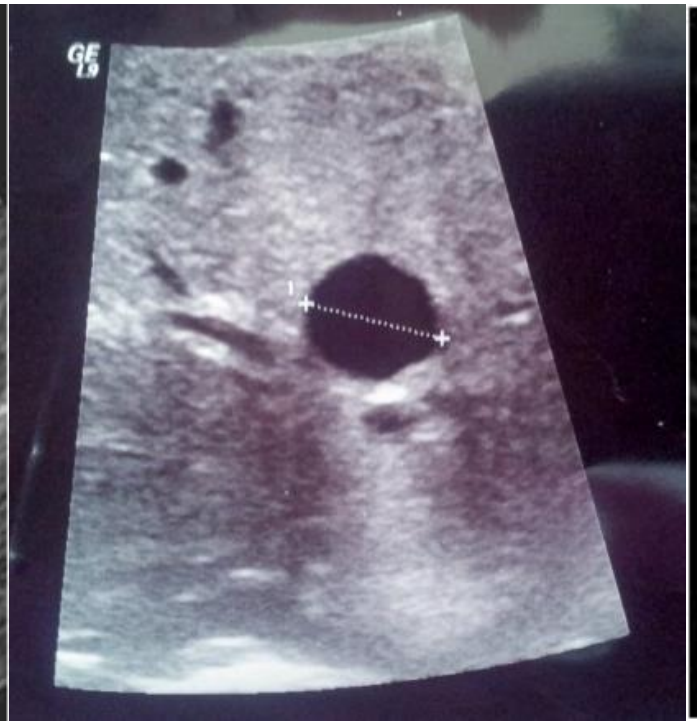
- Tam kan sayımı
- Endokrinolojik incelemeler: sT4, TSH, ACTH, Kortizol
- Enfeksiyonlar: İdrar ve kan kültürü, serolojik testler
- Metabolik testler
  - Alfa 1 antitripsin, Pi fenotip
  - Ter testi
  - İdrarda redüktan madde
  - İdrar organik asit
  - Tandem mass spektrometri
- Vertebra grafileri
- İleri metabolik testler
- Kas-deri biyopsileri
- Kranial MRI
- Kromozom analizi-Genetik testler
- Konsultasyonlar
  - Göz muayenesi
  - Kardiyoloji

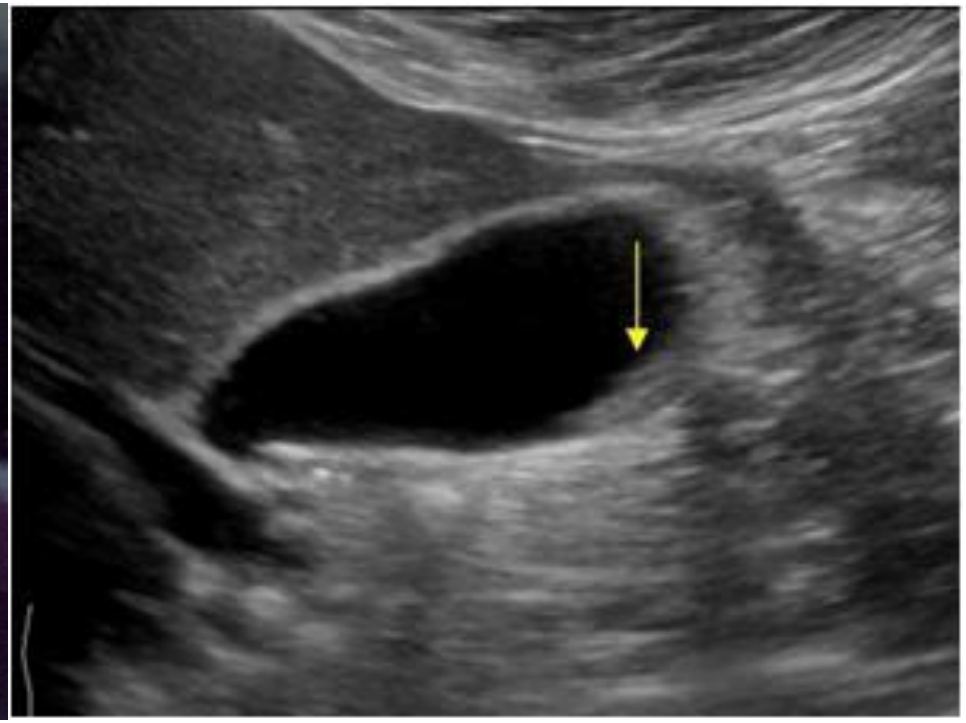
BAŞLANGIÇ TESTLER

- KCFT Paneli
- İdrar ve kan kültürleri
- İdrarda redüktan madde
- İdrar süksinilaseton
- Tandem mass spektrometri
- Abdominal ultrasonografi

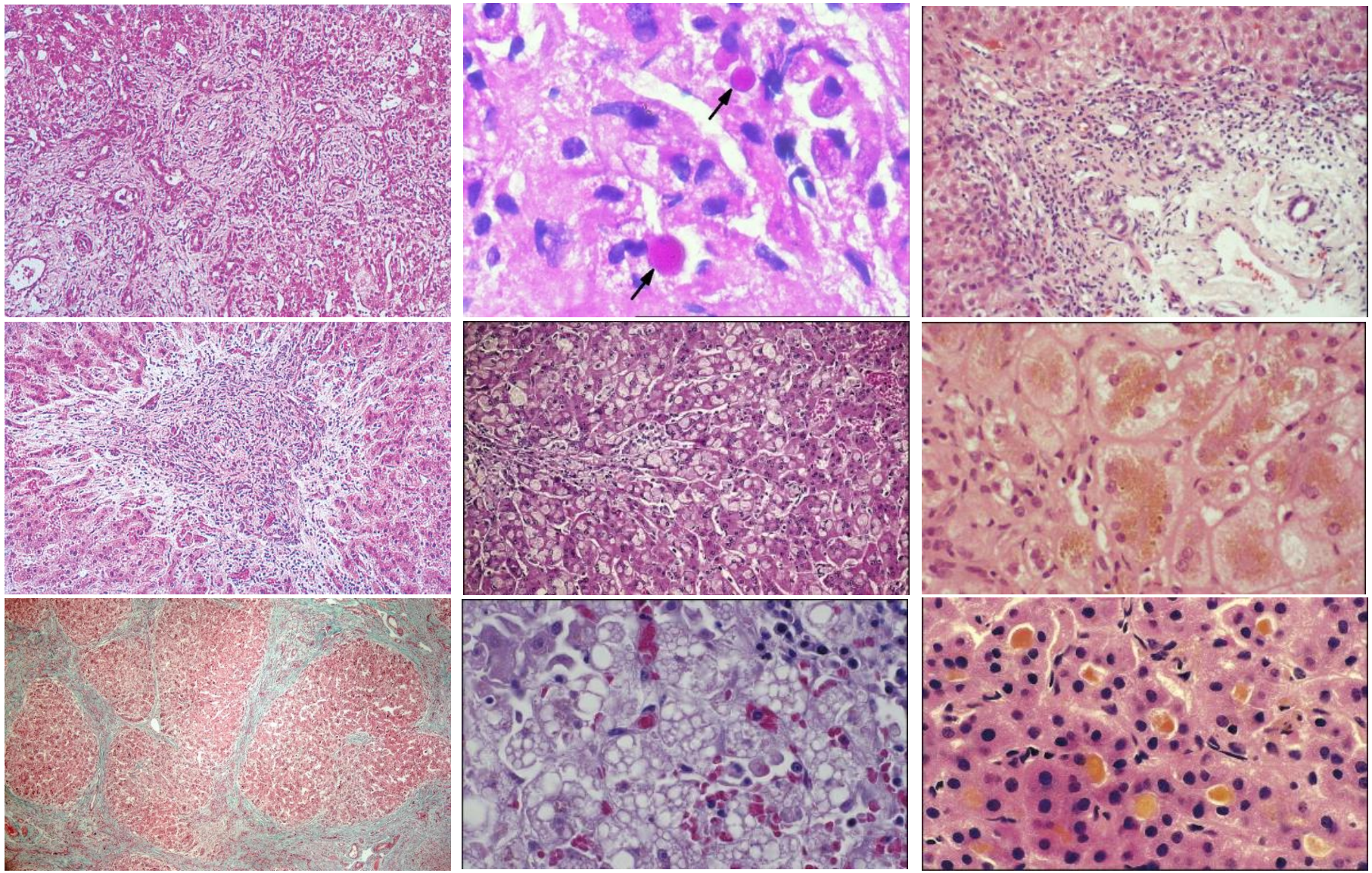
Ultrasonografi





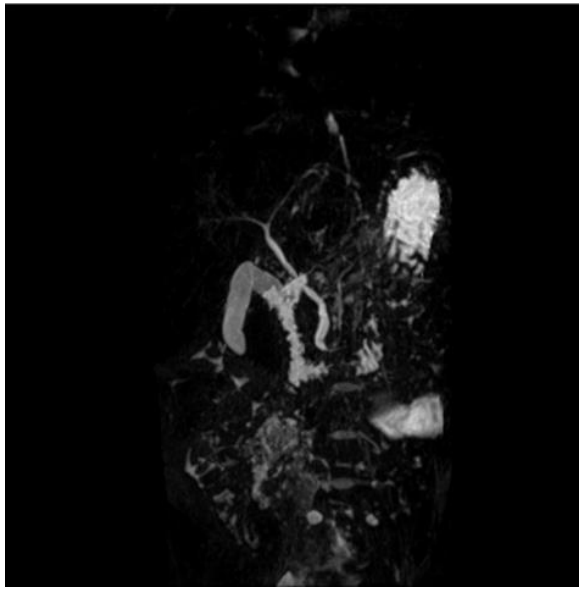


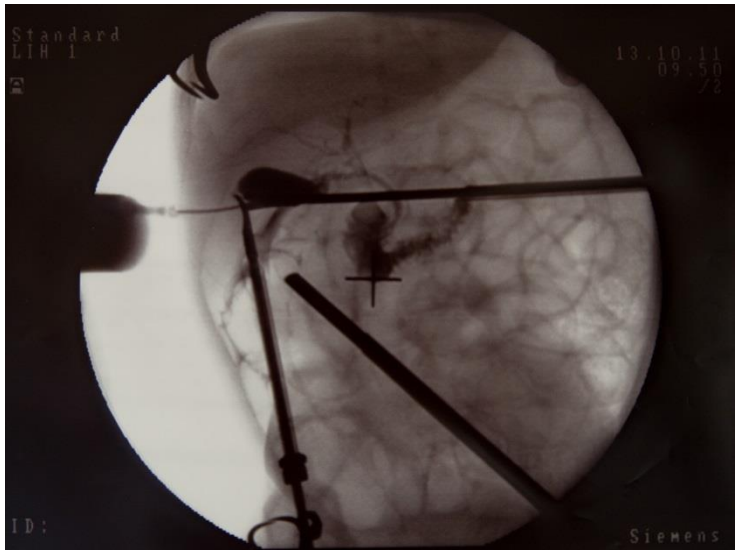
Karaciğer biyopsisi

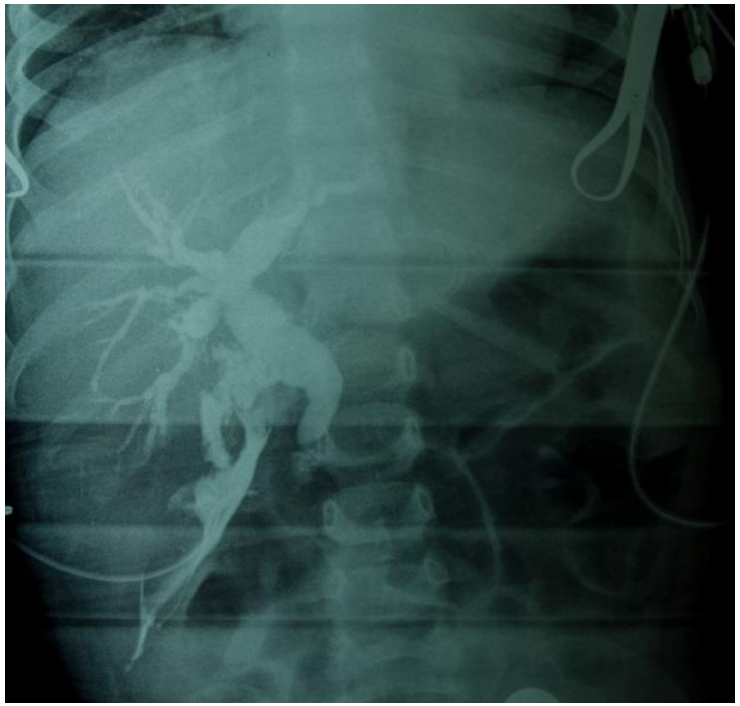


Kolanjiyografi

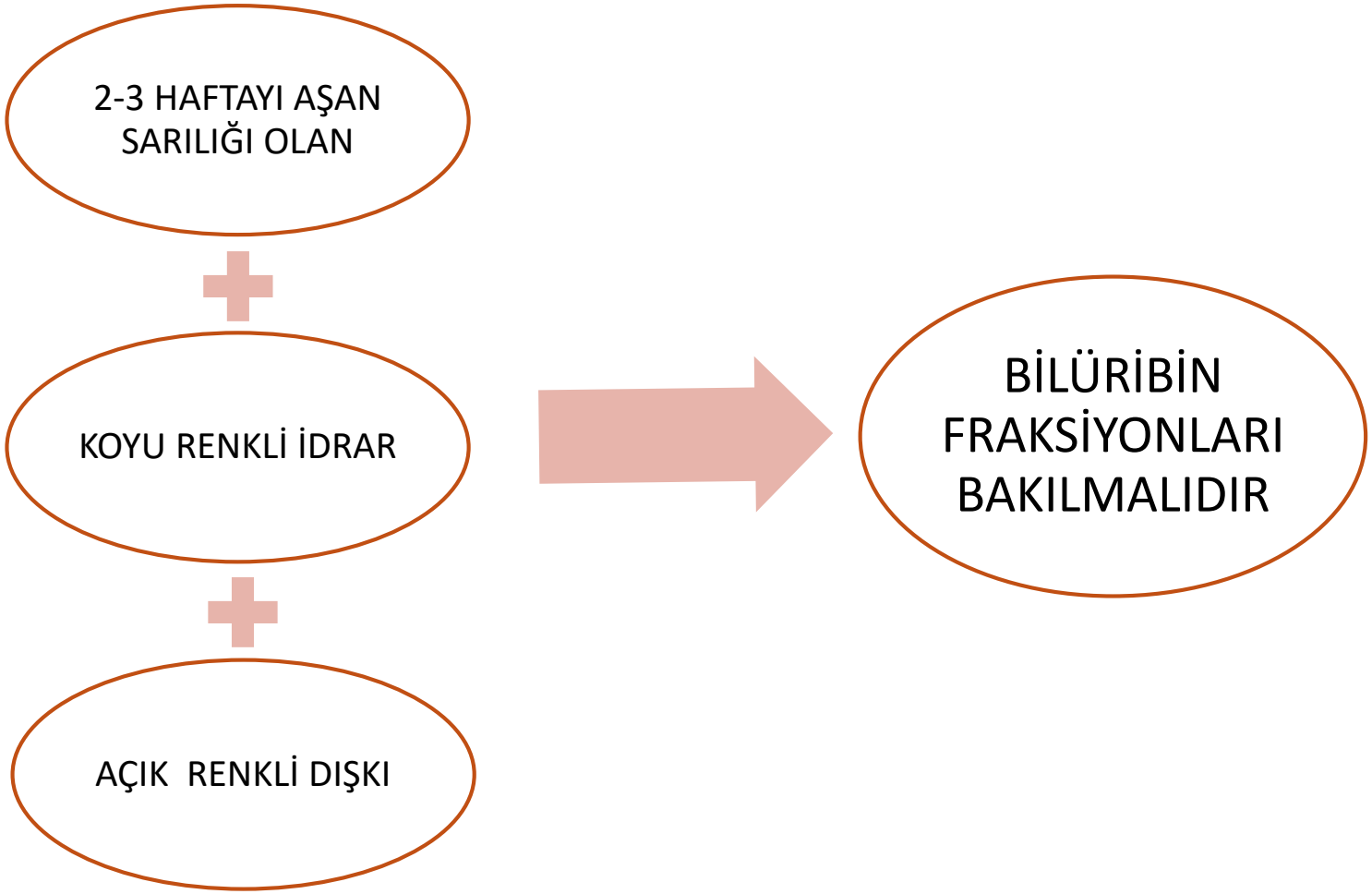
- MRCP
- ERCP
- İntraoperatif kolanjiyografi

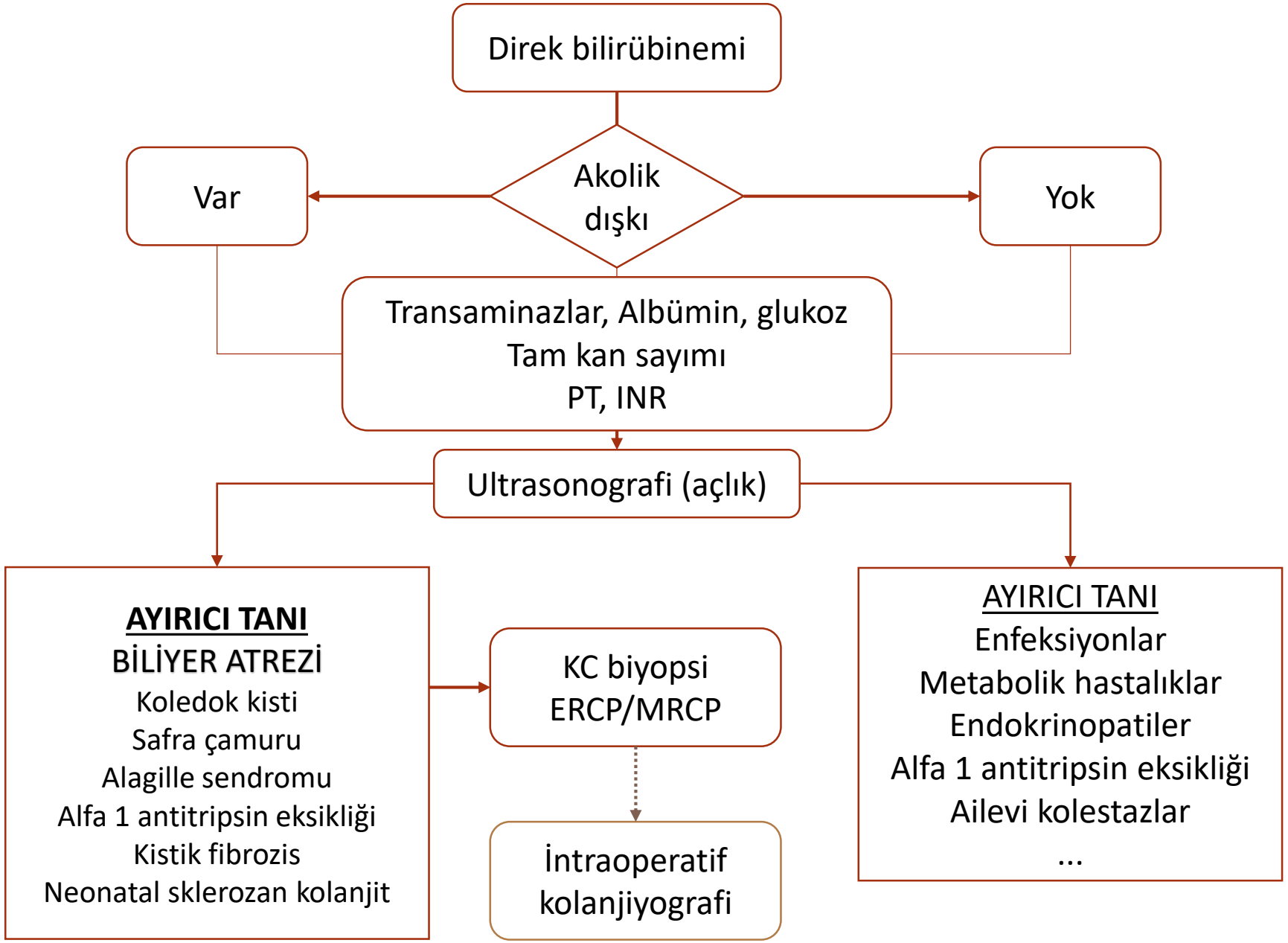






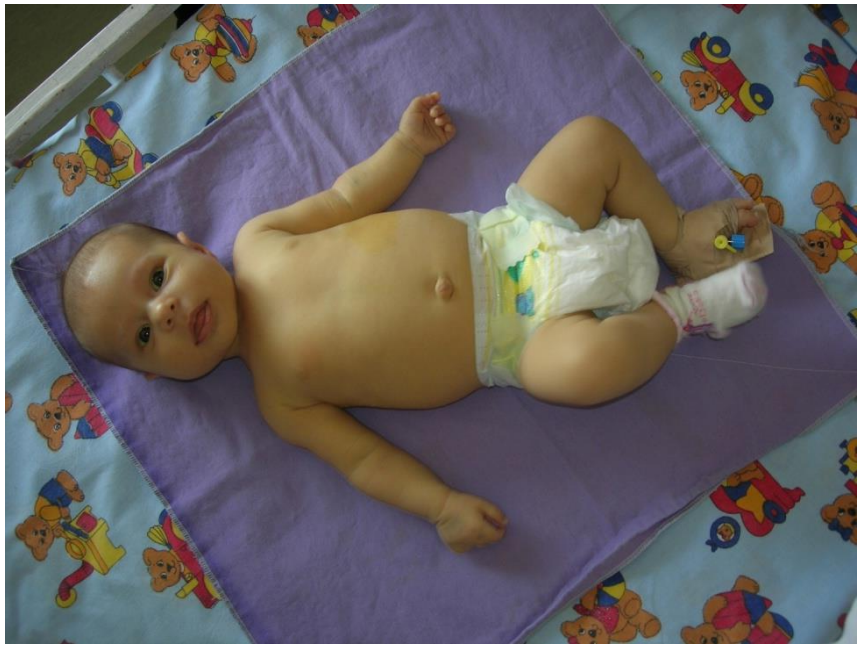
Tanı

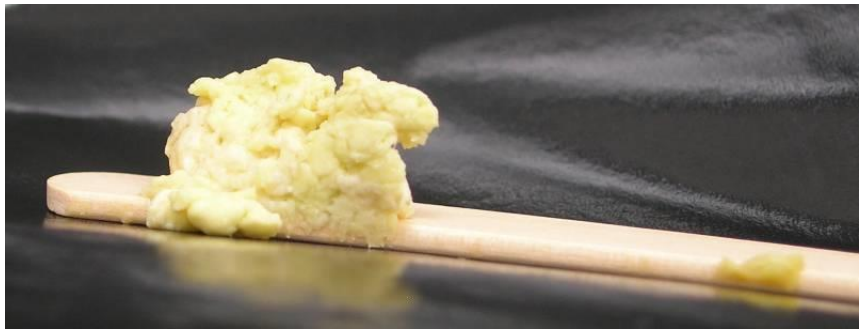

2-3 HAFTAYI AŞAN SARILIĞI OLAN
KOYU RENKLİ İDRAR
AÇIK RENKLİ DIŞKI
BİLÜRİBİN FRAKSİYONLARI BAKILMALIDIR

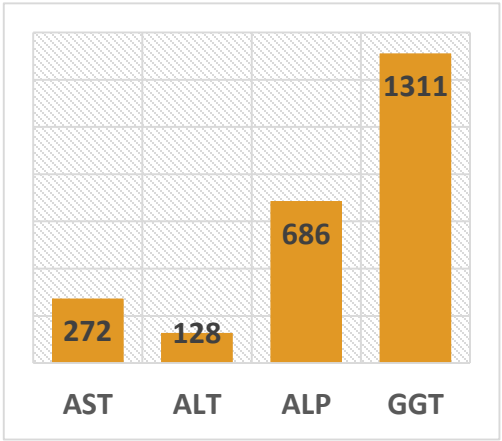

Direk bilirübinemi
Akolik dışkı
Var
Yok
Transaminazlar, Albümin, glukoz
Tam kan sayımı
PT, INR
Ultrasonografi (açlık)
AYIRICI TANI
BİLİYER ATREZİ
Koledok kisti
Safra çamuru
Alagille sendromu
Alfa 1 antitripsin eksikliği
Kistik fibrozis
Neonatal sklerozan kolanjit
KC biyopsi
ERCP/MRCP
İntraoperatif kolanjiyografi
AYIRICI TANI
Enfeksiyonlar
Metabolik hastalıklar
Endokrinopatiler
Alfa 1 antitripsin eksikliği
Ailevi kolestazlar
...

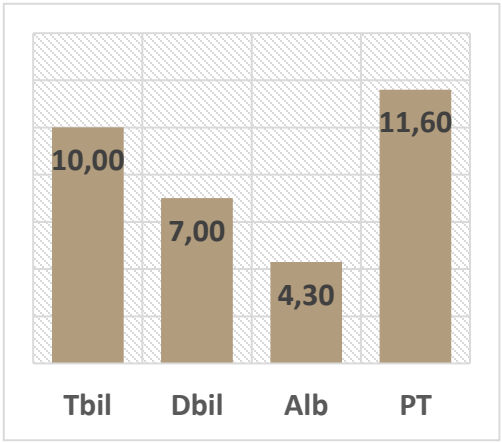
KRİTİK 6 HASTALIK

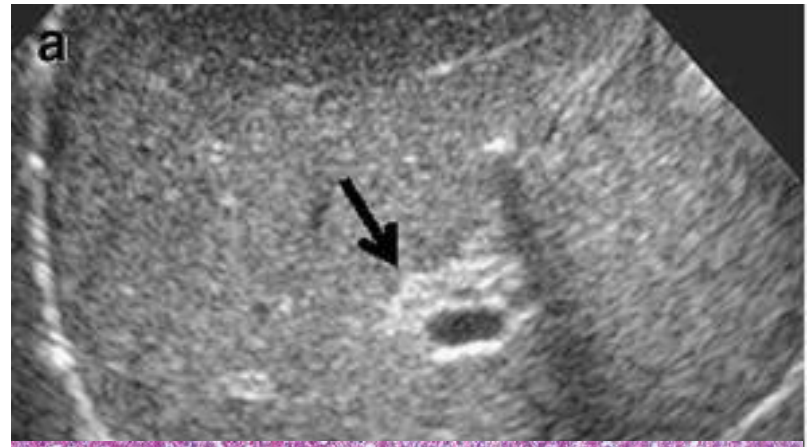
- BİLİER ATREZİ
- GALAKTOZEMİ
- TİROZİNEMİ
- HİPOTİROİDİZM
- SEPSİS
- UROSEPSİS

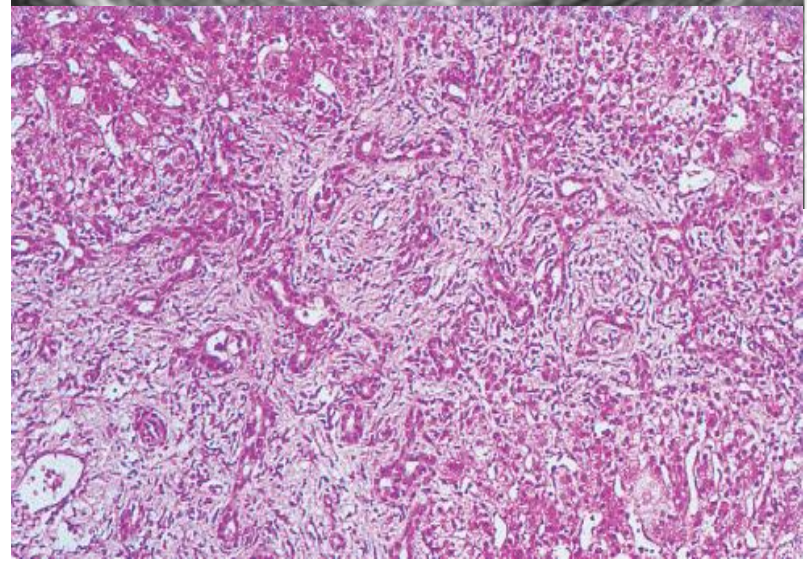








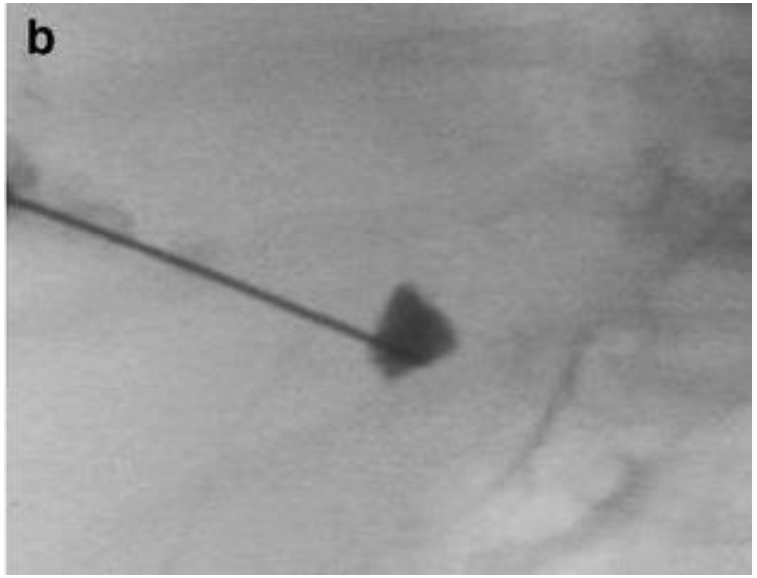


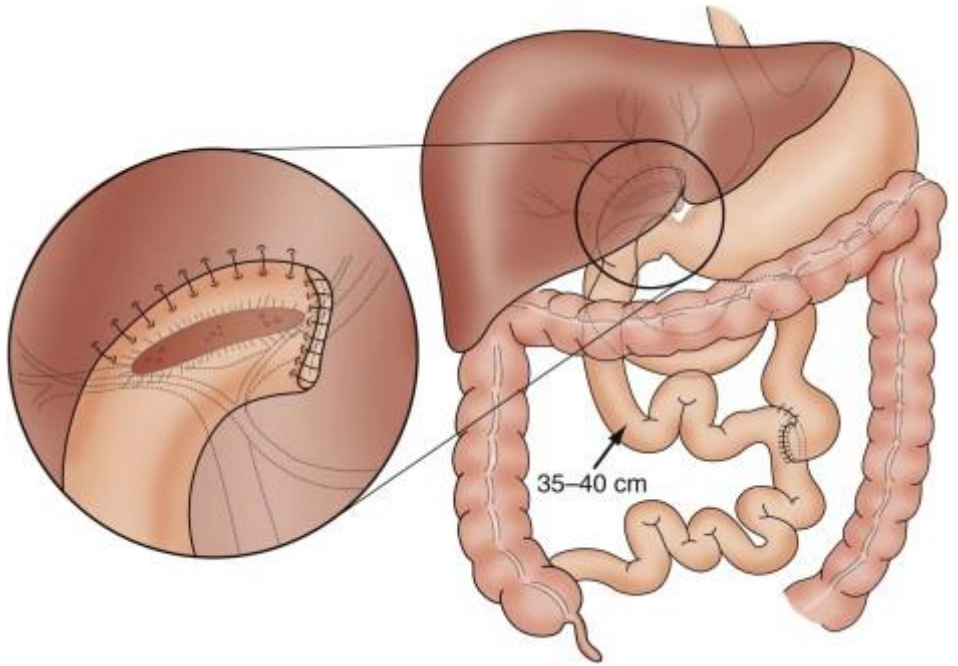


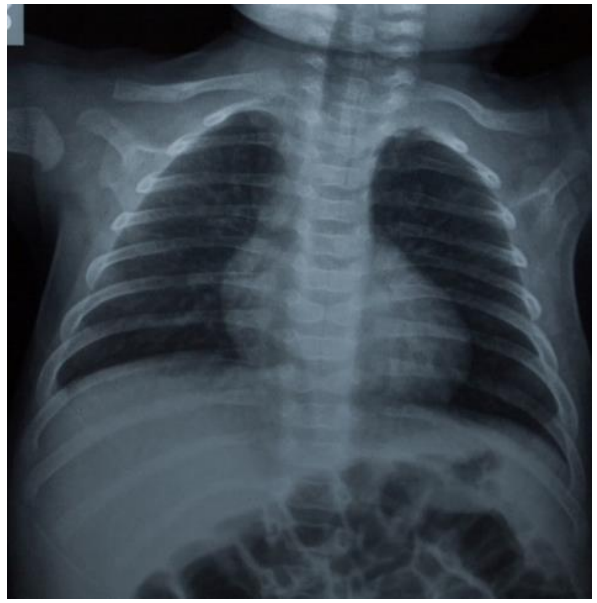
Biliyer atrezi

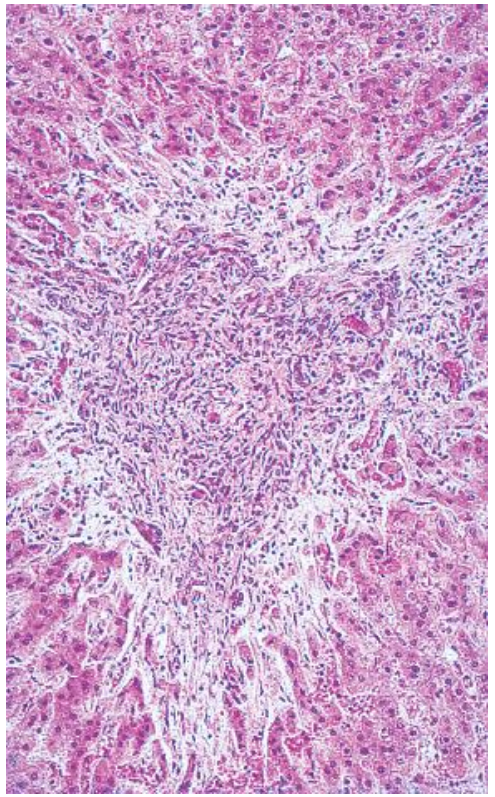
CERRAHİ

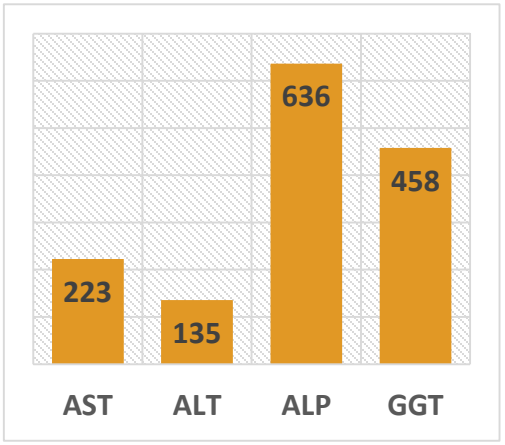
- İntraoperatif kolanjiogram
- Kasai portoenterostomi
- Çıkarılan ekstrahepatik biliyer materyalin patolojik incelemesi

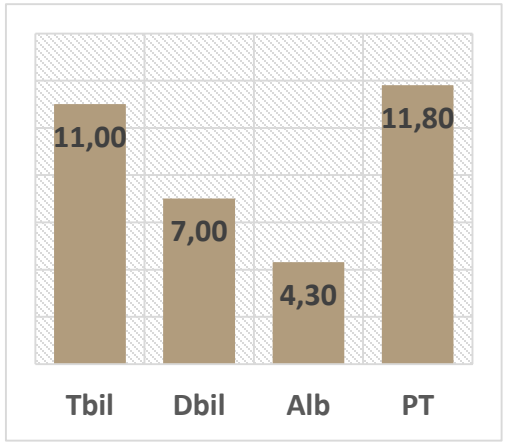




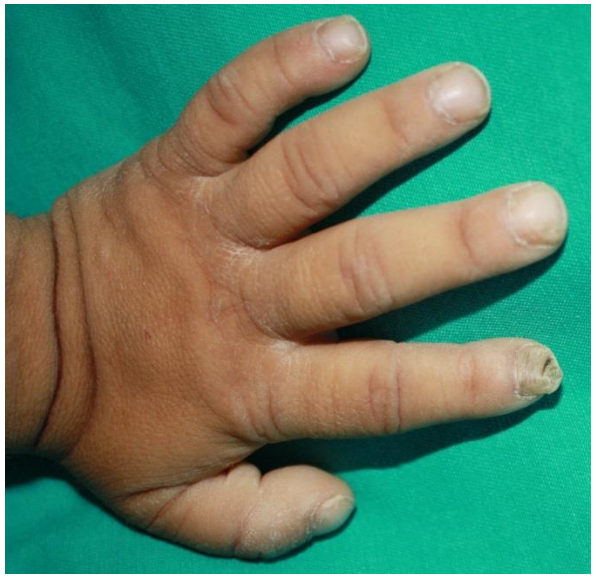


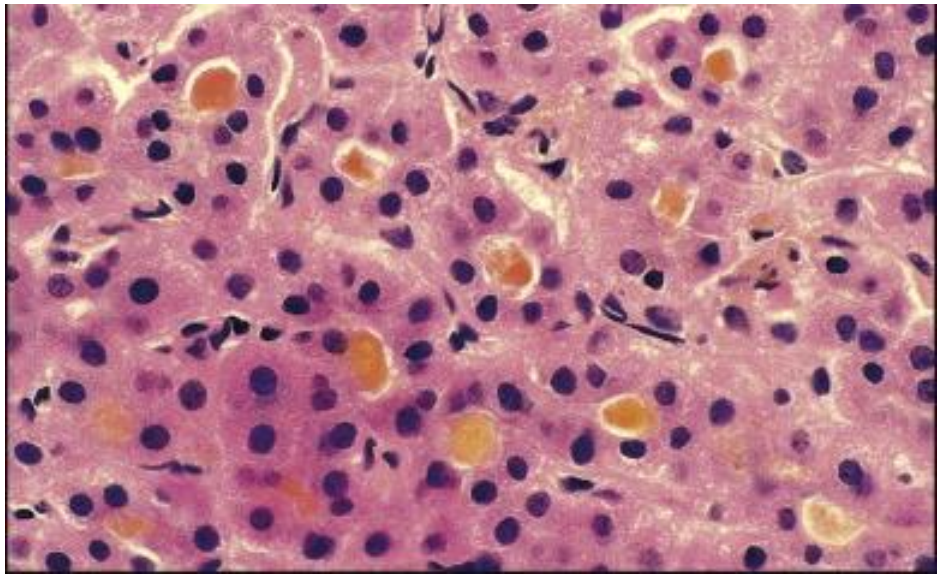




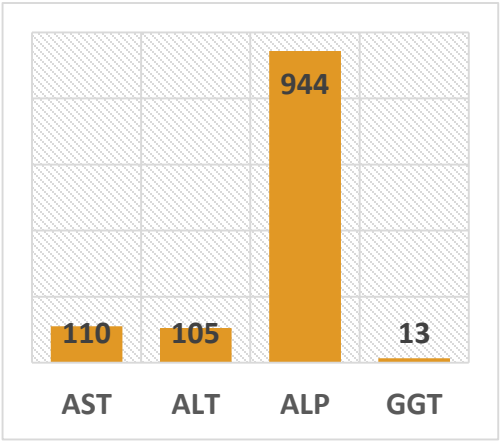


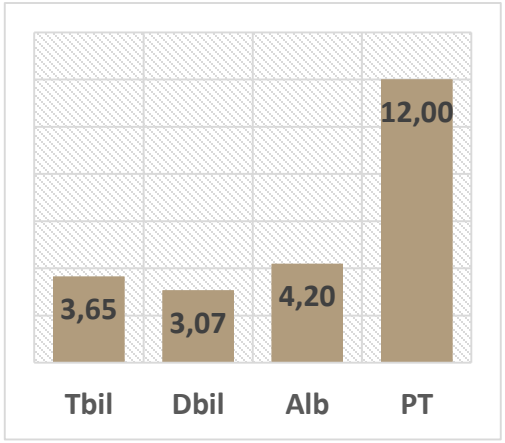
Alagille sendromu



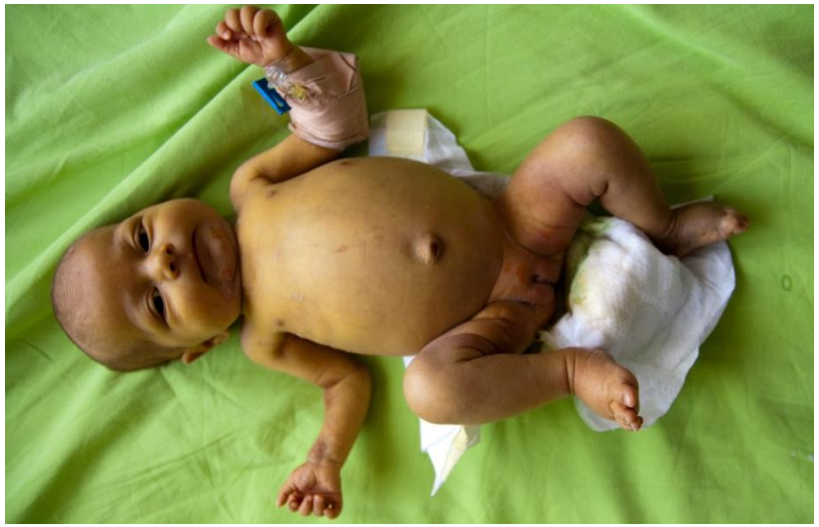


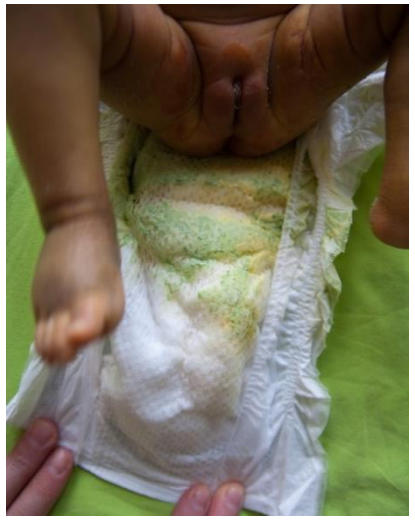
İshal
Akrabalık +

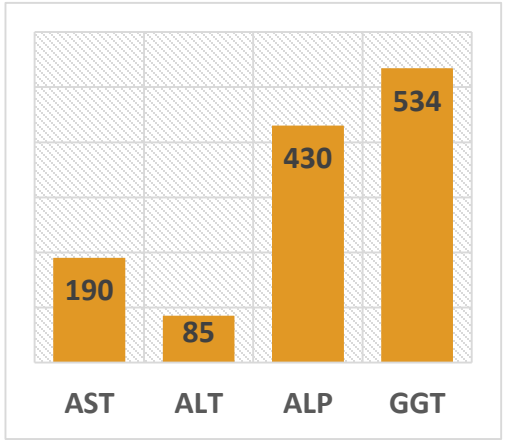


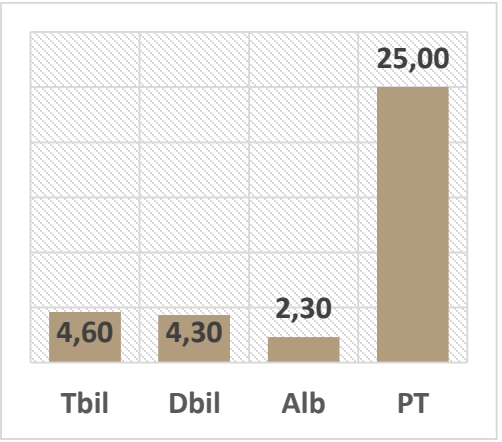


Progresif familial intrahepatik kolestaz (PFİK) Tip 1



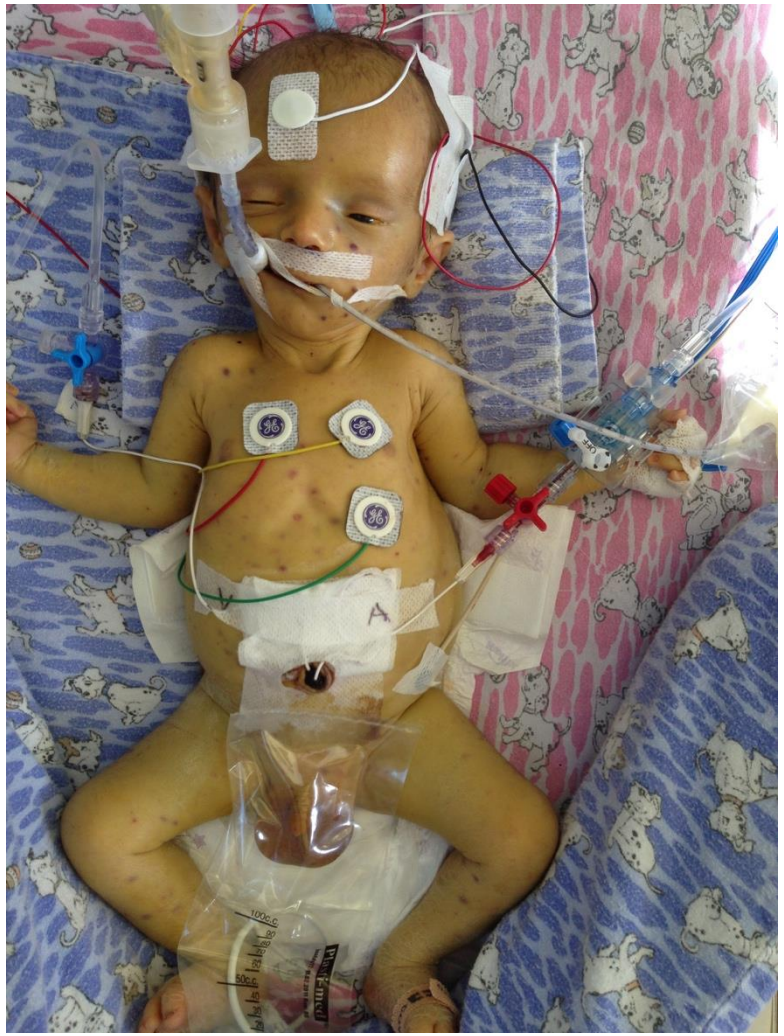


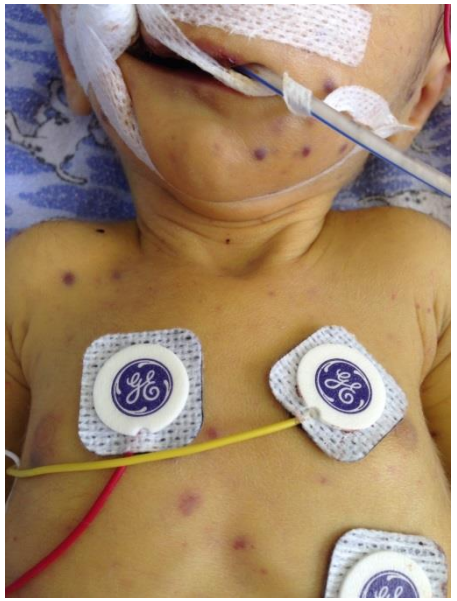




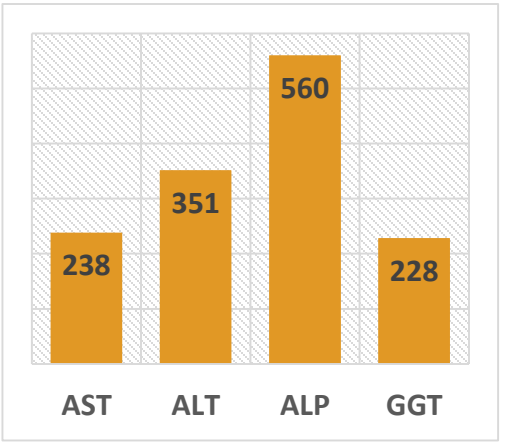
Hb: 6,5 g/dl
Rtc: % 5
Hiperkloremik metabolik alkaloz

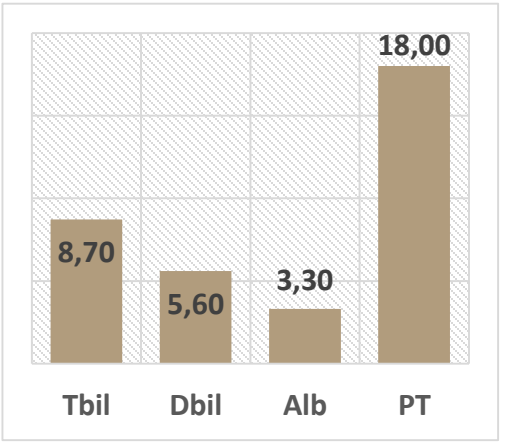
Kistik fibrozis



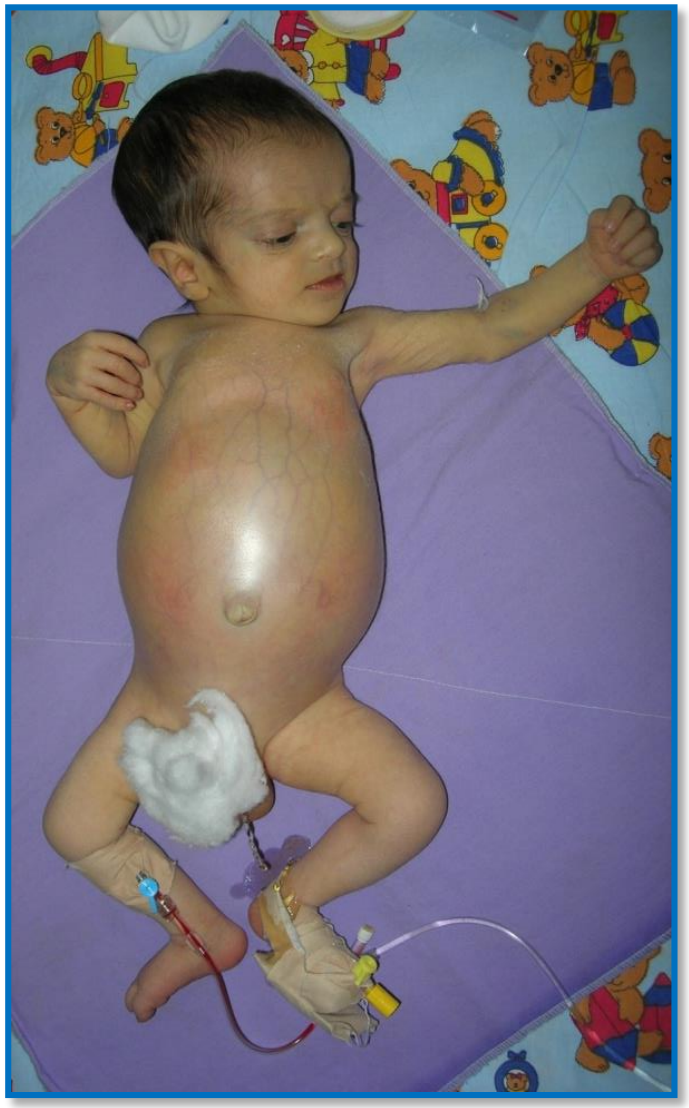


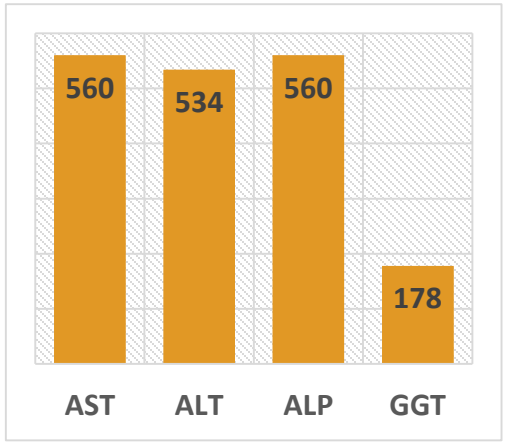
Düşük doğum ağırlığı
Koryoretinit
Mikrosefali

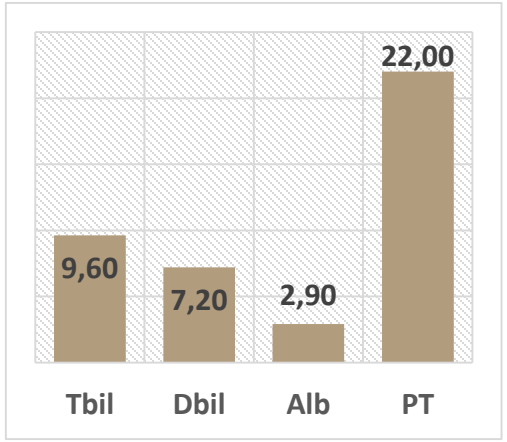




Konjenital CMV

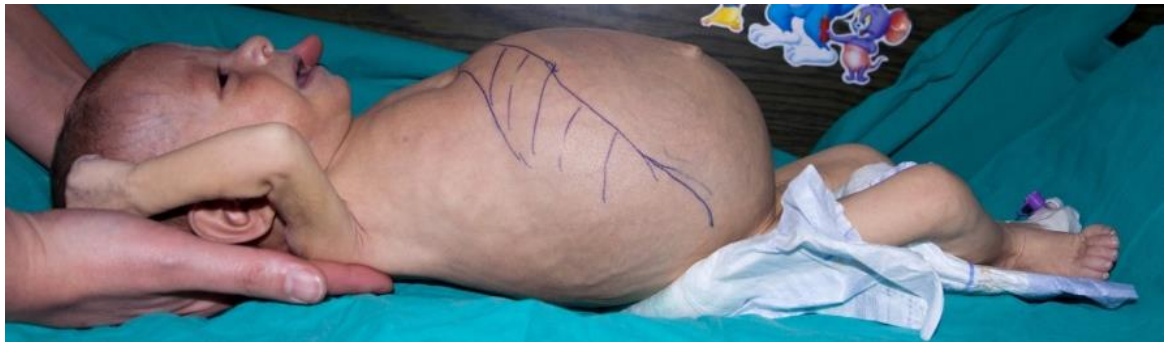


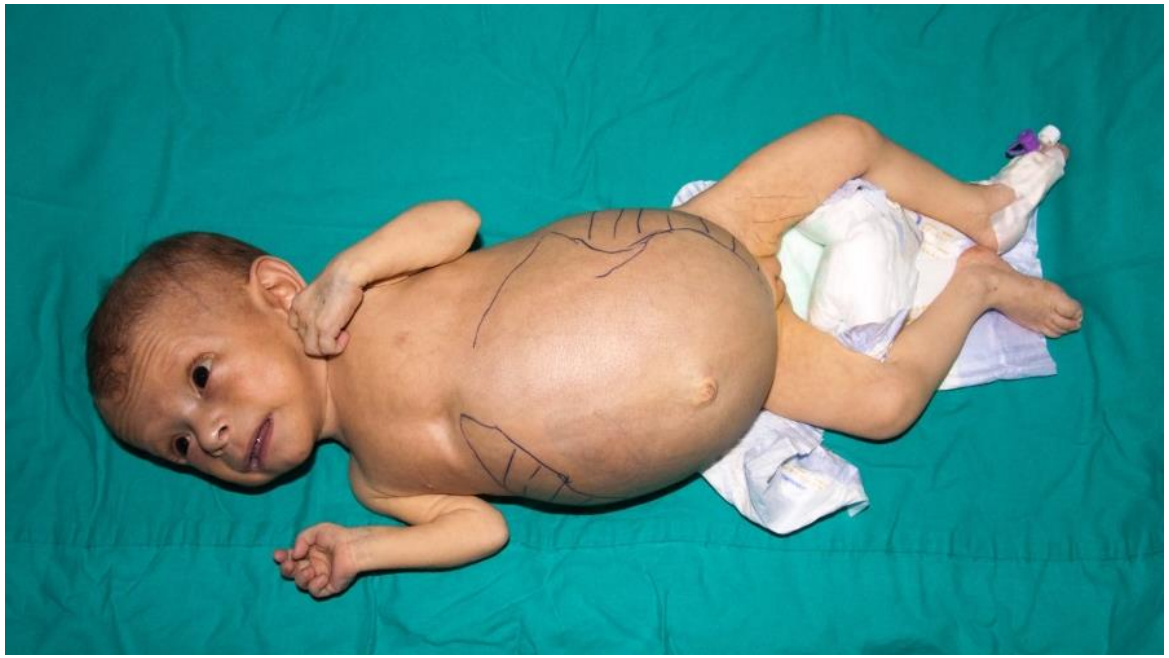


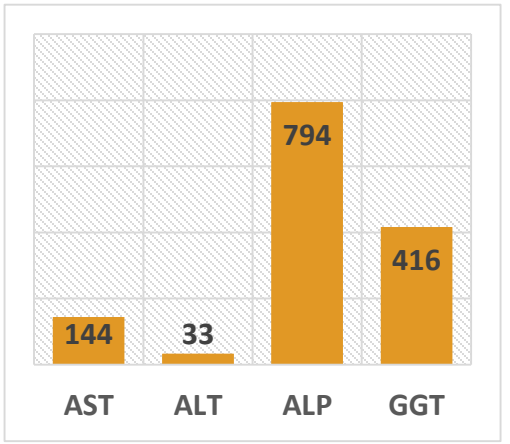


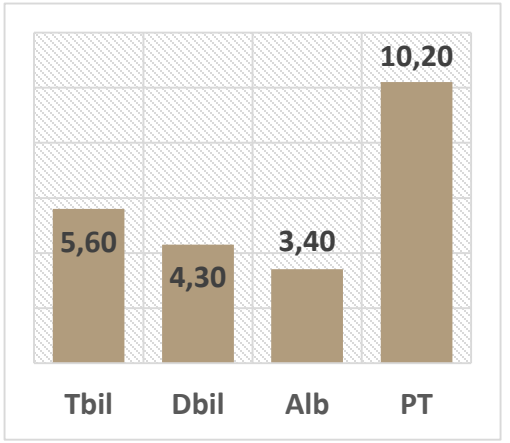
Katarakt
Kan kültürü: E. coli

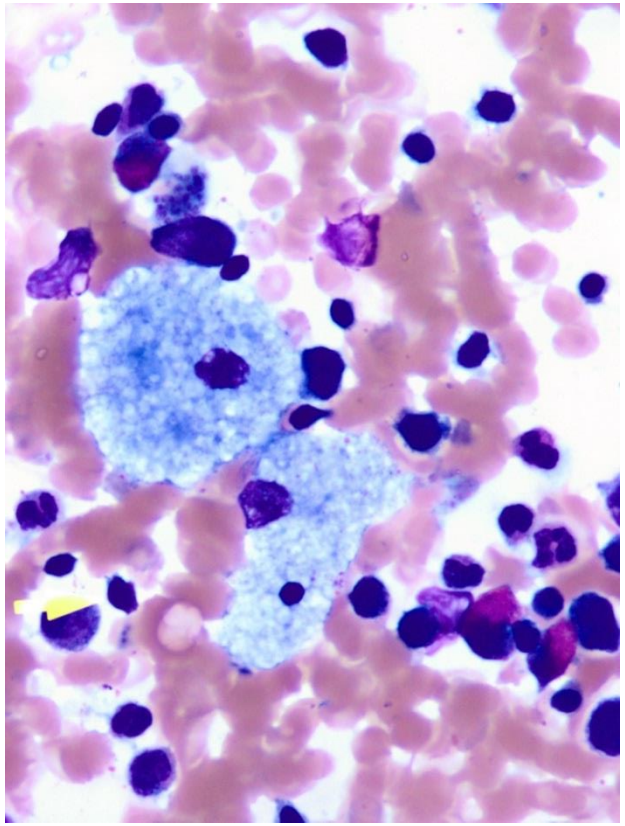
Galaktozemi



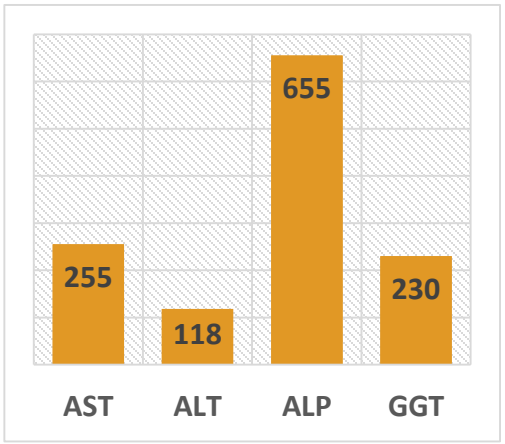


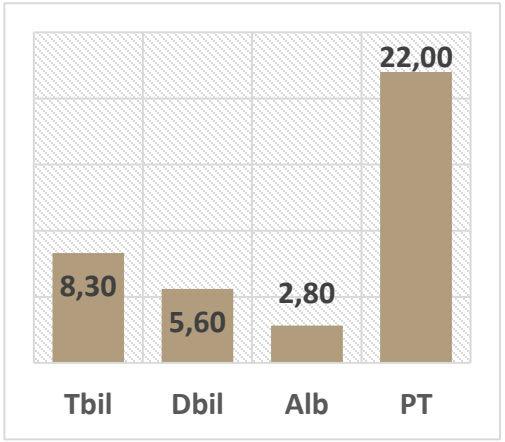


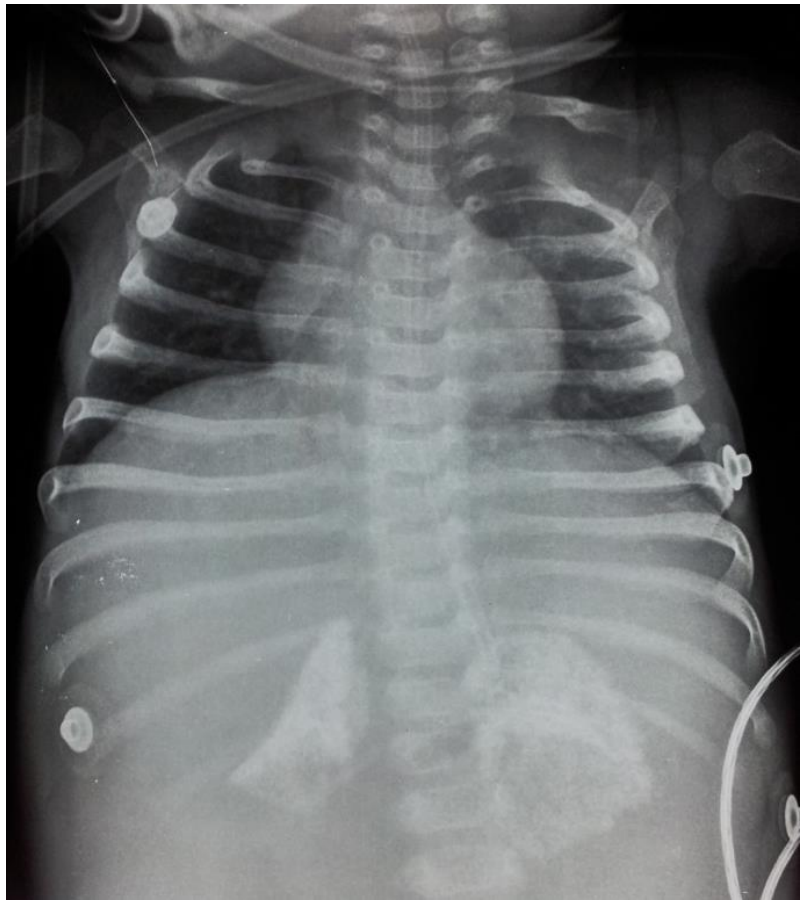




Niemann-Pick Tip C







Wolman hastalığı

Tedavi

Destek tedavi

- **Beslenme:** Anne sütü, MCT zengin mamalar
- **ADEK**
- **Kaşıntı:** UDCA, kolestiramin, rifampin, fenobarbital, naltrekson, ondansetron, cerrahi

Ursodeoksikolik asit

Büyüme-gelişme izlemi

Aşılar

Tedavi

Komplikasyonların tedavisi

Nedene yönelik tedaviler

- Laktozsuz mama
- Enzim tedavileri, NTBC
- Enfeksiyonların tedavisi
- Hormon replasmanı

Cerrahi

- Kasai portoenterostomisi
- Biliyer diversiyon
- Karaciğer nakli