Q: Polycythemia represents an abnormal elevation in the circulating RBC mass, increasing the risk of hyperviscosity, microcirculatory hypoperfusion and multisystem organ dysfunction. Of the following, the hematocrit threshold that best defines polycythemia is:
   A. 55%
   B. 60%
   C. 65%
   D. 70%
   E. 75%

A: C – 65%
http://neoreviews.aappublications.org/cgi/content/full/neoreviews;12/1/e20

Q: These two photos were taken on a 20 month infant born at 28 wks GA. What complication of his NICU course resulted in this finding?

A: Staphylococcus aureus septicemia

Q: Describe the finding and any associated abnormalities/syndromes, etc.

A: The picture shows coloboma on both irises. It can be associated with CHARGE, cat eye syndrome, Wilms Tumor-Aniridia association and multiple chromosomal anomalies (e.g., trisomy 13, del 13q, del 4p). The lesion may be limited to the iris (often the inferior part as shown at 6 o’clock), but it may reflect an underlying defect extending to the fundus/retina and optic nerve, so eye exam is warranted. In the latter case, vision may be severely impaired.

Q: Name this finding; with what syndrome is it most commonly associated; and how frequently is it seen in this syndrome?

A: Brushfield spots, seen in up to 95% of patients with Down syndrome
Q: What is the appropriate therapy for this abnormality? In what patient populations is it most commonly found?

A: Umbilical hernia. The most appropriate therapy is observation as incarceration is rare and most regress spontaneously; surgical repair may be indicated if still present at 3 years of age. More common in blacks, premature infants, and in patients with congenital hypothyroidism.

Q: Which of the graphs in the attached figure depicts total energy content per kg body weight?

A: B
Q: Which of the infant shown in Fig 20 is most likely to have the karyotype shown in (2 photos) Figure 4?

A: E
Q: How often is there a positive maternal history for the infectious disease shown in the attached photo?

A: < 50%

Q: Name the prominent features of the syndrome shown in the attached photo?

A: Cerebro-Hepato-Renal Syndrome; hypotonia, enlarged fontanel, defects of early brain development including pacitymicrogria, heterotopias/abnormal migration, mental retardation, hepatomegaly with dysgenesis and cirrhotic changes, albuminuria and glomerular cysts. The metabolic deficiency is a lack of DHAP-AT, a peroxisomal enzyme with a major role in glycerol ether lipid synthesis. AR inheritance with two chromosomal loci identified with mutations.
Q: What is the GA of the infant whose hand is shown in the attached photo?

A: 43 weeks (<40)

Q: Which of the infant shown in Fig 20 is most likely to have the karyotype shown in (2 photos) Figure 2?

A: B
Q: The etiology of the hand deformity shown in the attached photo is?

A: Multiple amniotic bands.

Q: A catheter is inserted into the umbilical vessel but it is difficult to determine whether the vessel is umbilical artery or vein. A pressure transducer is attached to the catheter and the pressure tracing shown in the attached figure is obtained. The pressure tracing is most consistent with location of the catheter tip in the:

A. aorta above the diaphragm,
B. portal vein system,
C. inferior vena cava above the diaphragm,
D. aorta below the diaphragm,
E. right ventricle.

A: C
Q: On the standard ventricular function graph shown in the attached figure, the dotted curves represent measured shifts from the solid curve. Identify the curves that correspond to decompensation and beta-blockade.

A: B, D

Q: Which of the infants shown in Fig. 20 is most likely to have the karyotype shown in Fig. 5?

A: A
**Q:** A 1.1 kg preterm infant is intubated successfully after several attempts. A nurse reports having difficulty inserting a nasogastric tube beyond 10cm, which is level with the diaphragm. A barium swallow is shown in the attached figure. Of the following the most likely diagnosis is:

A. esophageal atresia with distal TE fistula  
B. perforation of the proximal esophageal pouch  
C. traumatic perforation of the esophagus  
D. vascular ring  
E. esophageal web

**A:** C

**Q:** A BAER was performed on 65 very high risk newborn infants prior to discharge from the nursery in order to identify those at high risk for hearing loss. Each patient was re-evaluated at 3 years of age by an audiologist and the results are shown in the attached figure. The specificity of the BAER is:

A. 15%  
B. 52%  
C. 67%  
D. 85%  
E. 96%

**A:** E
Q: Which of the infant shown in Fig 20 is most likely to have the karyotype shown in Fig. 5?

A: D

Q: Name the syndrome most likely associated with these ear abnormalities and micrognathia.

A: Goldenhar Syndrome
**Q:** Diagnosis Please?

**A:** Syphilitic dactylitis

**Q:** With what syndrome/diagnosis is this finding commonly associated?

**A:** Periventricular calcifications seen with congenital CMV.
Q: Identify these lesions.

A: Bohn nodules

Q: Please identify, and what should you do with an infant presenting with this lesion?

A: Epulis—usually arises in the area of the incisor. No specific therapy required, as they are benign and usually regress spontaneous—unless it is large enough to cause feeding difficulties. Then, an ENT or oral surgeon can address it. Some use the term epulis to refer to ANY tumor of the gingiva.
Q: Syndrome?

A: Treacher Collins syndrome

Q: What do you think was the presentation of this baby? Any associated concerns?

A: Breech presentation
Q: Identify this component of the ECMO circuit and list which types are available for use, including the criteria for choice.

A: Membrane oxygenation. The two types available are heparin bonded (Carmeda is what we have always used) and non-heparin bonded (Avecor). The advantage to the heparin bonded circuit is that you do not need to fully systematically heparinize the patient and can thereby avoid some potential bleeding problems. This has traditionally been the circuit of first choice in our neonatal patients. The Avecor circuit has a longer life, though, and you can avoid complications of clot formation, oxygenator failure and hemolysis.

Q: Diagnosis please...

A: Turner’s syndrome
Q: Diagnosis please and life expectancy?

A: This is an x-ray of thanatophoric dysplasia, a generally lethal form of short limb dwarfism. The name comes from the Greek term thanatophoric, meaning “death bringing,” since most patients die shortly after birth—due in large part to the small thoracic cage and restrictive respiratory insufficiency. Transmission is autosomal dominant; therefore, all cases represent new mutations. There are two types. Type I is characterized by the curved long bones and very flat vertebral bodies. Type II is characterized by straight femora and cloverleaf skull (Kleeblattschadel anomaly). If patients live, they have profound developmental delays and severe FTT. Prenatal diagnosis can be made, but be sure about the techniques. Just ask Ex about the delivery he was asked to attend to pronounce an infant with this diagnosis made prenatally and how the infant lived.
Q: What is the differential diagnosis for this presenting symptom?

A: Macroglossia, Beckwith-Weidemann syndrome

Q: What is the diagnosis, including most commonly associated conditions/syndromes?

A: Cherry red spot: Tay-Sachs; Neimann-Pick; generalized gangliosidosis syndrome Type I
Q: Which of the graphs in the attached figure depicts total energy content per kg body weight?

A: B

Q: A catheter is inserted into the umbilical vessel but it is difficult to determine whether the vessel is umbilical artery or vein. A pressure transducer is attached to the catheter and the pressure tracing shown in the attached figure is obtained. The pressure tracing is most consistent with location of the catheter tip in the:

A. aorta above the diaphragm,
B. portal vein system,
C. inferior vena cava above the diaphragm,
D. aorta below the diaphragm,
E. right ventricle.

A: C
Q: On the standard ventricular function graph shown in the attached figure, the dotted curves represent measured shifts form the solid curve. Identify the curves that correspond to decompensation and beta-blockade.

A: B, D

Q: Which of the infants shown in Fig. 20 is most likely to have the karyotype shown in Fig. 2?

A: A
Q: A BAER was performed on 65 very high risk newborn infants prior to discharge from the nursery in order to identify those at high risk for hearing loss. Each patient was re-evaluated at 3 years of age by an audiologist and the results are shown in the attached figure. The specificity of the BAER is:

\[
\begin{array}{c|cc}
   & \text{Yes} & \text{No} \\
\hline
\text{BAER} & \text{Abnormal} & 4 & 9 & 13 \\
            & \text{Normal} & 2 & 50 & 52 \\
            &     & 6 & 59 & 65 \\
\end{array}
\]

A. 15%
B. 52%
C. 67%
D. 85%
E. 96%.

A: E

Q: Which of the infant shown in Fig 20 is most likely to have the karyotype shown in Fig. 5?

A: D
Q: Which of the curves shown in the figure is most closely associated with the hemoglobin beta chain?

A: C

Q: Syndrome?

A: Rubenstein-Taybi syndrome

No Question, just an interesting case from the New England Journal of Medicine
Q: Identify the types of ECMO pumps and indicated use:

A: Roller pump and vortex/centrifugal pump

Q: Diagnose Please:

A: Fetal Alcohol Syndrome