**NOTE 2**

**PATIENT 1001**

**Date: 4/25/22**

**Patient Summary**

Patient is a term infant with Coffin Siris, Dandy walker malformation, aortic coarct on PGE, and concern for tracheomalacia admitted for airway evaluation and cardiac intervention planning.

**Presenting History**

Male born at 38w5d via induced vaginal delivery to a 30 y/o G1P0 mother with prenatal screens as documented below. Maternal history unremarkable. Maternal medications include PNV and bASA. Pregnancy notable for GBS positive, fetus with FGR 2%,  Dandy-Walker malformation, c/f partial agenesis of the corpus callosum, bilateral rocker-bottom feet and webbing of toes, abdominal and hepatic cyst, mandibular protrusion, cardiac defects (borderline hypoplastic aortic valve, elongated LVOT, PFO and PDA but no mVSD), filling defect of bladder (atypical but nonspecific finding).  
    
Mother was admitted to L&D for "IOL for FGR 2% and Dandy-Walker malformation, partial agenesis of the corpus callosum, bilateral rocker-bottom feet, abdominal and hepatic cyst . Received: None. Antibiotics during labor: penicillin. Labor notable for: None.  
   
Delivery History  
Per neonatal delivery note "OB/GYN Attending at delivery was Grace John Kor, MD, PhD  
   
Cord clamping was delayed by 30 seconds.  
   
The infant emerged weak following induced vaginal delivery with poor cry and poor tone for gestational age. He was brought to the DR warmer and dried, stimulated, and bulb suctioned. Other interventions included: nasopharyngeal suctioning, SpO2 monitoring and positive pressure ventilation until ~ 3 mins then infant transitioned to CPAP. HR initially was 60-100 and increased during delivery room observation and interventions. Birth weight was 3060 g (6 lb 11.9 oz)". Of note, infant voided in DR.  
   
Maternal Labs  
GBS: +  
  
   
Lab Results  
Component    Value    Date  
     ABO    O    03/12/2022 blood type  
     RHTYPE    Positive    03/12/2022   
     ABSCRN    Negative    03/12/2022 antibody screen  
     RUBTITR    immune    10/01/2021 rubella   
     HBSAG    Neg    10/01/2021 hep b surface antigen  
     HCVAB    negative    10/01/2021 hep c antibody  
     RPR    non reactive    10/01/2021 syphilis  
     HIVABS    Neg    10/01/2021 HIV antibody  
     NGONCLT    Neg    10/19/2021 niceria ghon-- culture  
     CHLAMCLT    Neg    10/19/2021 chlamydia  
     VARIGG    immune    10/01/2021 varicella technically a positive resul  
----------------------------------------------------------------------------------------------------  
Genetic Screening:  
low risk NIPT, cell free DNA and carrier screening  
  
Prenatal ECHO  
  
US OB  Biophysical Profile With Measurements and Doppler  
Performed: 3/1/2022 at  4:18 PM  
Accession Number: E33691752  
Reason For Exam  
  
Dandy-Walker malformation, partial agenesis of the corpus callosum, bilateral rocker-bottom feet, abdominal and hepatic cysts; Suspected/Known Fetal Abnormality  
       
Impression  
Best GA (EDC): 37w0d, EDC: 3 - 22 – 2022 (estimated)  
 Fetus 1:  
Estimated Weight: 2307 gm +/- 392 gm [ 5 lbs and 1 oz] Weight Percentile for Gestational Age: 2 %ile  
   
Fetal Anatomy:  
HEAD: \*\*Abnormal, Known Dandy walker malformation. Enlarged cisterna magna, absent vermis and splayed cerebellum. CSP not visualized  
SPINE: \*\*Unevaluable  
FACE: \*\*Unevaluable  
NECK/THORAX: Normal  
HEART: \*\*Abnormal, Cardiomegaly, Right side appears larger than the left. Difficult to fully evaluate due to fetal position and late gestational age.  
STOMACH: Small, posterior fluid-filled structure seen, likely stomach  
KIDNEYS: Normal  
BLADDER: \*\*Not Seen  
ABDOMEN (OTHER): Prominent spleen. Hepatic cyst not definitely seen.  
VENTRAL WALL: \*\*Unevaluable  
EXTREMITIES: Right upper present,  Left upper present,  \*\*Right lower abnormal,  \*\*Left lower abnormal, Bilateral rocker bottom feet  
UMBILICAL CORD VESSELS: 3-vessel cord  
   
Fetal Measurements:  
 BPD  : 85 mm  
 OFD  : 108 mm  
 HC   : 313 mm  
 AD   : 97 mm  
 AC   : 303 mm ( 4 %ile)  
 FL   : 64 mm ( 2 SD below mean)  
 FL/AC: 0.21 (normal range 0.20-0.24)  
 HC/AC: 1.03 (normal range 0.9 - 1.1)  
   
 Basis for GA:  
 BPDc: 35w2d  
 BPD : 35w2d  
 FL  : 33w4d  
 EDC : 37w0d --- (EDC: 3 - 22 - 2022)  
   
Maternal History  
Information for the patient's mother  
History reviewed. No pertinent past medical history.  
Past Surgical History:  
Procedure    Laterality    Date  
•    HERNIA REPAIR            
•    INGUINAL HERNIA REPAIR            
     2014 and 2017  
  
#    Outcome    Date    GA    Lbr Len/2nd    Weight    Sex    Delivery    Anes    PTL    Lv  
1    Term    03/13/22    38w5d    / 01:40    3060 g    M    Vag-Spont    EPI         LIV  
  
-------------------------------------------------------------------------------------------------------------  
   
ROS:  Negative except as noted above  
   
**Prior NICU course:**  
**Access:** PICC DOL 3-present. EPIV DOL 12- 32. Hx of TPA to PICC on 4/21- successful.  
   
**CV:‎**Infant admitted to NICU with diagnosis of possible coarctation of aorta and started on PGE. PGE was discontinued on DOL 7 after normal echo and infant subsequently had systolic BP gradient of 15-20 with weak pulses. Echo repeated and again demonstrated a coarctation of the aorta with no PDA. PGE restarted at this time. PGE briefly discontinued on 3/29 per cardiology in order to obtain anatomically accurate cardiac CT. Cardiac CT done on 03/31, shows atypical "napkin ring" coarctation of the aorta per Dr. Dorn. Given results PGE restarted on 4/1. (please see multiple separate echo reports for details). Plan to transfer patient for dynamic bronchoscopy to help decide to proceed with tracheostomy or a pexy procedure. This will ultimately guide decision to proceed with a stent in the cardiac lab or proceed with cardiac surgery and timing of procedures  
   
**Resp**:‎ Infant required intubation and conventional ventilation following admission to NICU. Multiple failed extubation attempts, ORL bedside scope reassuring with no further workup necessary unless infant is unable to wean off respriatory support in future (per Beth Landon, MD).  Received airway Dex course on 3/28-3/29 and 4/4-4/5 in preparation for extubation. Infant successfully extubated to HFNC 6L on 4/5. Required escalation of support to HFNC 10L on 4/8 iso of increasing number of significant desaturation events. Continues to require frequent suctioning for clear secretions without improvement. Per discussion with ORL (Beth Landon, MD) 4/11. team does not wish to visit at this time but will reassess pending cardiology's plan. Dr Sean Brone (pulmonary) consulted 4/13, Atrovent started per his recommendation. Infant had significant ABD events requiring PPV overnight 4/12-4/13. Placed back on CPAP 8 DOL 4/14 with fewer severe events noted thereon after. CPAP interface changed to RAM to help with agitation.  
 -------------------------------------------------------------------------------------------------------------------------------  
**FEN/GI**:‎ Hx of NPO while c/f coarctation. Infant cleared to start enteral feeds of MM only on 4/3 by cardiology following the cardiac CT. Advanced to full volume feeds of MM/DM at 150 ml/k/d on 4/12, and has tolerated without issue. Continues on Vit D supplements. Currently NPO for planned procedure.  
   
**Heme**: Hyperbilirubinemia risk factors: none.  Maternal blood type O+ and infant O+ direct coombs negative. Peak bili on 3/19, did not require phototherapy.  Initial HCT 49, plts 208K. History of direct hyperbilirubinemia iso of prolonged PN, infant subsequently transitioned to SMOF (through 4/8) with improvement noted in follow up labs. Most recent GGT on 4/24 improving, hct 32.3.  
   
**ID**:‎ Screening CBC and BC sent iso GBS + mother and clinical illness. BCx NGTD: final. CBC reassuring. Amp/Gent x 48 hours complete. MSSA+ 4/16. Completed course of mupirocin on 4/20 and has had subsequent negative culture. 4/21 persistently febrile with peak temperature 38.6. Blood and urine cultures obtained and nafcillin and gentamicin were started for 48-hour course. Blood culture NGTD x4 days. Urine Culture negative final.  
 ------------------------------------------------------------------------------------------------------------------------------------  
**GI**: Fetal US noted cyst on liver and in abdomen.  Abd US on DOL 0 confirms 10 mm cyst left lobe of liver. Also noted distended rectum, normal variant, neurologic or hirschsprung's. Unable to appreciated abd cyst but may be obstructed by rectum. Radiology recommends repeating study once infant has stooled.  Repeat 3/15 shows liver cyst and 4 mm submucosal nodule in the bladder. Repeat 3/28 shows partially distended bladder with a 3 mm submucosal nodule protruding into the bladder lumen along the posterior upper to mid bladder wall, as seen previously. Continued follow-up is recommended. Repeat US should be obtained around 04/29/22  
   
**GU:**Unable to palpate testes on initial exam. US of scrotum noted undescended left intraperitoneal testis, right testis not visible.  May be obstructed by rectum. Recommends repeating study once infant has stooled several times. Of note, bilateral inguinal hernias noted on US. Repeat US 3/15 shows undescended intraperitoneal right testis, undescended left testis positioned high in inguinal canal and bilateral inguinal hernias; presently inguinal hernias have not been palpable.  
   
**Neuro**: Known fetal anomalies including Dandy Walker Malformation and agenesis of corpus collosum. HUS on DOL 0 confirms Dandy Walker Malformation and dysgenesis of corpus collosum. MRI done on 3/17, with similar findings from prenatal scans and no hydrocephalus. Neurology unable to give prognosis, requests genetic results prior to discussing. Hx of requiring IV precedex and Versed for sedation. Transitioned to PO ativan + clonidine, agitation improved with increased clonidine dose. In preparation for OR on 4/25, precedex restarted at 0.2 mcg/kg/hr and Ativan 0.07 mg/kg IV Q6 hours.  
 --------------------------------------------------------------------------------------------  
**Genetics:**Mother met with genetics counselor prior to delivery.  Cord blood sent for microarray.  Genetics consulted on 3/14, spoke with family. They requested CDT (Congenital Disorders of Glycosylation) and Karyotype, which have resulted as normal. Parent consent and approval obtained for whole genome sequencing.  May be able to obtain Whole Exome from cord blood. Microarray reveals "normal XY". Genetics received FOB sample on 3/27. Whole Exome report received on 4/12:  Heterozygous for a De Novo Likely Pathogenic Variant in AMARCA4; (Coffin-Siris syndrome 4). Heterozygous for a Variant of Uncertain Significance in CHD7.  Genetics team spoke with mother at bedside on 4/13.  
   
**Endo:**Abnormal DOL 30 NBS. TSH is 14.4, (RR <10). Total T4 = 7.3. Repeat values on 3/2 showing values within normal range (TSH 4.55 and FT4 1.1)  
   
**Social/Family**. Planning on transfer on 04/25 for 11:30AM Dynamic Bronchoscopy.  
   
Discharge Disposition: See ICOR note.  
   
RHCM: Routine Health Care Maintenance Prior to Discharge:  
[X] Red reflex- pale bilaterally.  
[X] Hip exam  
[x] Hepatitis B Vaccine 3/13 given  
[ ] Hearing Screen:   on  .  
[X] CCHD screen: Multiple echocardiograms, confirmed CHD.   
[ ] Car Seat Test    
[ ] Circumcision (if parents request and consent)  
[ ] Identify PCP  
[ ] Newborn Screens   
24 HOL Date Sent:3/14  Result: In range  
DOL 14 Date Sent: 3/27  Result: C0 (Acycarnitines), Leucine (Leucine) out of range, repeat NBS    
DOL 30 NBS sent early on Date Sent: 4/10  Result: TSH is 14.4, RR <10. Total T4 = 7.3.  
DOL 60 Date Sent:  Result:   
Discharge Date Sent:  Result:   
-----------------------------------------------------------------------------------------  
**Post-op Addendum 4/25/22:**  
ORL: right vocal cord difficult to visualize but was briefly visualized, mild edema of supraglottis, tarchea and bronchi without malacia on nondynamic exam. Grade 1 view, intubated with 3.5 uncuffed ETT.  
Pulm: Per report, noted to have 75% collapse on right with cough, 50% collapse on left, no malacia distally, no significant secretions, no BAL sent. No indication for pexy.  
Anesthesia: Remained on precedex infusion. Received remifentanyl and propofol. Continued PGE infusion. Intubated with 3.5 uncuffed ETT by ORL, +leak, received airway dexamethasone x1. Initially noted to have diminished breath sounds on the left, ETT in adequate position at 9cm at the gum, aeration improved.  
Plan: see below  
   
**Plan**  
*CV:*Hemodynamically stable. Napkin ring coarctation of the aorta, currently planning for surgical repair, continue PGE 0.05.  
Access: PICC (3/16-) will obtain XR to verify tip placement   
   
*Resp*: History of stridorous breathing and desaturation events, previously on bCPAP 8, concerning for tracheomalacia. DLB 4/25 without significant malacia or secretions, no indication for pexy. Admitted intubated, s/p airway dex x1, leak present during DLB, consider additional airway dex as needed. XR and gas on admission. Wean to extubation, extubate to CPAP. Continue atrovent nebs. Pulmonary following, will discuss plan from pulmonary perspective.  
   
*FEN*: Previously feeding MM/DM TF150cc/kg/day. Currently NPO on IVF with lytes. Plan to resume NG feeds once extubated. Continue vitamin D. Monitor I/O, feeding tolerance, weights.  
   
*GI/Bili*: History of direct hyperbili i/s/o prolonged PN, follow LFTs/GGT qMonday. Last AUS 3/15 with liver cyst and submucosal nodule in bladder, plan to repeat AUS 4/29, monitor.  
   
*GU:*Undescended testes high in inguinal canal and bilateral inguinal hernias confirmed on US 3/15, follow clinically, will need surgical evaluation for hernias after cardiac repair.  
----------------------------------------------------------------------------------  
*Heme*: Obtain HCT, monitor.  
   
*ID*: Recent sepsis evaluation 4/21 with naf/gent x48h, blood and urine cultures NGTD. No active concerns, monitor.  
   
*Neuro*: Dandy walker malformation and agenesis of corpus callosum confirmed on MRI 3/17, neurology following. Daily HC. Continue precedex infusion, wean as needed. Continue clonidine and ativan.  
   
*Genetics:*Congenital disorders of glycosylation and karyotype normal. Microarray XY male. WES - heterozygous for de novo variant in AMARCA4, coffin siris syndrome, heterozygous for a VUS in CHD7. Genetics following. Will need follow up with cancer dept regarding screening for tumors given CSS.  
  
*Endo:*DOL 30 abnormal TFTs, labs 4/17 reassuring (TSH 4.5, FT4 1.1).  
  
*Other*: PACT following, last meeting 4/5.  
   
*Social*: Updated on admission. Portuguese speaking. Last family meeting 4/19.   
   
*Disposition*: pending extubation to baseline respiratory support, consider retrotransfer to HOSP 1 while awaiting upcoming cardiac repair  
   
*RHCM:*  
- Newborn screening: 3/14 normal, DOL 30 abnormal TFTs, labs 4/17 reassuring, repeat per protocol, next DOL 60  
- Hepatitis B Vaccination: 3/13  
- Other Vaccinations: will need  
- CCHD: multiple echos, not indicated  
- Hearing screen: will need  
- Car seat testing: will need  
- Circumcision: will inquire  
   
PCP: will identify and update  
  
I agree with the details of the note above. I participated in the management of the patient, performed the service and was physically present during the key portions of the service when performed by the fellow, Dr. Yuan Enners  
  
Lance Paul, MD