Case Study 2: Pierre Robin Sequence

NURS 6035 Practicum I

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Case Study 2: Pierre Robin Syndrome

Preliminary Information

Selection of Case

I had the opportunity to spend some of my clinical hours this semester with Dr. Drummond-Borg at Cook Children’s genetics clinic. I chose this case because I was not familiar with Pierre-Robin sequence (PRS) or with its associated genetic conditions. I think that it is an interesting case, and I wanted to share the information that I learned with my peers.

Type and Number of Encounters

This is N.E.’s second visit to the genetics clinic. The patient was seen one-month ago (September 14, 2009) for an initial evaluation of PRS. Today, October 17, 2009, the patient returns to review genetic test results.

Insurance

This patient is on the state-funded Medicaid, PCCM plan.

Subjective

Patient Profile

Five month-old Caucasian female, N.E., presents to the genetics clinic today with her biologic mother.

Background Information

CC. Patient born with PRS with cleft palate. Returning to genetics clinic today to review genetic lab testing.
HPI. Patient was initially seen in genetics clinic on 09/14/09 for physical examination in order to determine if there is a genetic causation related to the PRS.

PMH.

Birth. N.E. was born to a Gravida 3, Para 1, AB 1, 27-year old who reports no problems with the pregnancy. There was no exposure to medications, alcohol, recreational drugs, or tobacco. Mom had been previously evaluated for infertility because it took six years to conceive N.E. Mom reports that she has “had failure with ovulation” and her husband “has a very low sperm count.” Delivery was at 38 weeks gestation in local hospital in Gainesville, Texas.

Birth weight: 7 pounds 8.9 ounces (25-50th percentile).

Length: 51 cm (50th percentile).

Head: 33.3 cm (10-25th percentile).

Apgars: 8 at one minute: 9 at five minutes.

Labs: Unavailable

N.E. was noted to have a cleft palate and a small chin. There were some respiratory difficulties. She was transferred to Cook Children’s Medical Center for further evaluation, and within a few days she was discharged to home. Patient had plastic surgeon consult with Dr. Obaid prior to discharge.

One month. Having difficulty sucking and swallowing. Parents meet with “pediatric physical therapist and cleft palate specialist to make sure that we are using the Haberman and pigeon nipples correctly.”
Two-three months. N.E. continues to have difficulty sucking and swallowing formula. She continues to choke on saliva and has poor weight gain. She is also having difficulties with sleep apnea and difficulty maintaining a patent airway. She was noted to have an airway obstruction from a sleep study performed on 07/15/2009. The surgical procedure, myringotomy, was performed and pressure equalization (PE) tubes were inserted on 07/24/2009. A computerized tomography of the face (07/31/2009) showed a soft tissue cleft palate. N.E. had a mandibular distraction procedure performed on 08/04/2009. Following the jaw distraction, N.E. demonstrated persistent feeding problems. A swallow study was done which showed laryngeal penetration with thin, nectar consistency liquids present. A G-button was inserted on 08/26/2009.

Four months. N.E. has initial visit with Dr. Kukolich (Cook Children’s geneticist) on 09/14/09. Diagnosis of PRS confirmed. Genetics testing ordered by Dr. Kukolich at this time to determine if PRS is a component of a genetic syndrome.

Medications. Poly-Vi-Sol with iron infant vitamin supplement drops, 1.0 ml added to formula inserted through G-button daily.

Family History.

Mom. Denies smoking cigarettes, use of illegal substances, or consumption of alcoholic beverages (currently and during pregnancy). States that she is "in good health." Mom does not work outside of the home. She has a high school diploma and has completed some college courses.

Dad. Denies smoking cigarettes. Has a history of recreational drug use approximately 10 years ago. Denies current use. Usually consumes one
alcoholic beverage ("usually beer") on weeknights and might have two drinks on the weekend. Mother states that "he is in good health also," but states that he does not go to the doctor for routine physical examinations. Dad is employed full-time at a local concrete company. He has a high school diploma.

**Half-brother.** Patient has 8-year old half-brother from mom’s first marriage. He is “healthy” and he is currently seeing a physician for suspected attention deficit hyperactivity disorder.

**Additional family information.** Patient lives with biologic mother, father, and half-brother. Family resides in private home in Gainesville. Family had Blue Cross Blue Shield health insurance at the time that N.E. was born, however, two months ago the family had to cancel this policy due to financial constraints. They are currently receiving state funded insurance. All other extended maternal and paternal family members are reported to be in “good health.” There are no reported genetic syndromes on either side of the family (see Appendix for pedigree).

**Social History.** Besides her immediate family, patient has extended family (aunts, uncles, cousins, grandparents) that live nearby. The extended family spends time together on a weekly basis. Grandparents babysit on occasion so that parents can go out alone. Family is also very involved in their local church where patient attends weekly worship services. Patient does not participate in church nursery activities due to the risk of contracting infection or illness. Parents do not attend any support groups, and since patient has been born, they have been unable to attend their young couples’ Sunday school class.
Review of systems.

**Head/Neck.** Holds head up well. Turns side-to-side. Trachea midline. Neck is supple without masses or lymphadenopathy.

**Skin.** No rashes or birthmarks. Occasional diaper rash that resolves with the use of OTC diaper cream.

**Eyes.** Eyes don’t “cross” anymore. Has patient had an examination with the ophthalmologist? Mom does not recall a referral for an eye doctor appointment. Mom states, “Her vision seems normal.”

**Ears.** Has had one ear infection since birth. No infections since PE tubes inserted. No drainage from canals. Hearing is “normal.”

**Nose.** No problems with congestion or rhinorrhea. No respiratory distress noted since the surgery. No snoring or apnea spells noted.

**Oropharynx.** Mother notes that N.E. has mandibular hypoplasia and a cleft palate. “Drools a lot.” Parents attempt to feed her a small amount of baby food by spoon. N.E. enjoys the flavor but will not eat more than a spoonful. She will not suck on a nipple and only chews on it. Meets with physical therapist on a weekly basis. Patient is slow to make progress. Plastic surgeon plans to close cleft palate when patient is 12-months old.

**Cardiac.** Denies cyanosis. No heart murmurs detected by general pediatrician or any other specialist visit.

**Pulmonary.** Occasional loose, productive cough. Patient is able to clear secretions “better” during the past few weeks. Mom does not have to suction
patient. No hemoptosis, wheezing, or respiratory distress noted since surgery in August.

Gl. No problems with G-button. Receives all feedings and medications through G-button. Mom administers daily feedings based on calorie and fluid requirements established by nutritionist. No constipation noted. Has two loose stools daily. No hematochezia or hemorrhoids reported.

GU. Voids every 2-3 hours. No blood, discharge, or foul odor noted.

Neuro. Babbles, but does not say any consonants or words. Laughs and responds to family members, especially to half-brother. Mom does not think that there is any apparent neurologic damage, but she is unsure at this time. Has appointment with pediatric neurologist next month.

Musculoskeletal. Rolls from stomach to back. Sits unsupported. Moves arms and legs bilaterally. Reaches for objects and tries to put them into her mouth. Not crawling, however, she “scoots” and rotates in circles when placed on abdomen. Does not rock on hands and knees. Puts feet into mouth when lying on her back.

Growth/Development. Mom verbalizes that N.E. is “slower” in her development than other children. She is hopeful that she will catch up with the help of physical and occupational therapy. Since N.E. has recently been changed to Medicaid health insurance, she is not currently enrolled in Early Childhood Intervention (ECI).

Objective

Physical Examination
General. N.E. is a well groomed, five month-old Caucasian female who has bilateral metal distraction devices in place in her lower jaw.

Vital Signs.

Weight. 6.22 kg, 13.68 pounds (10-25th percentile)

Height. 67 cm (90th percentile)

Head. 41 cm (25th percentile)

Inner canthal distance. 2.5 cm (WNL)

Outer canthal distance. 7 cm (WNL)

Head/Neck. Head appears normocephalic. Symmetrical, anterior fontanel 3 cm., soft, flat, and pulsatile. Forehead is slightly prominent. Hair is brown color, fine, and evenly dispersed on scalp. Chin is now pulled into normal position and the distraction apparatus is intact bilaterally. Neck appears normal. There are no masses noted.

Skin. Shows no abnormal areas of hyperpigmentation or hypopigmentation.

Eyes. Eyes are slightly deep set. Solera, irides, and fundi are normal. PERRL bilaterally. No strabismus noted. Patient tracks light appropriately for age. No ptosis or epicanthal folds noted.

Ears. Ears are cupped. Both ears measure 4.4 cm. No ear protrusions or unusual structure or markings. Canals pink without discharge or excoriations. PE tubes intact and tympanic membranes are translucent, pearly, with landmarks noted bilaterally.

Nose. Nose is small, patent, symmetric, and centrally positioned on face. Turbonates are pink. No discharge or erythema noted.
Oropharynx. Philtrum is well formed. Lips are normal. Mouth appears normal. Palate shows a large posterior cleft. Tongue appears normal. There are no teeth present.

Cardiac. Heart sounds are normal and no murmur appreciated. All pulses are present, strong, and equal.

Pulmonary. Chest symmetric. Nipples are normal. Lungs clear to auscultation.

GI. Soft with bowel sounds present. No organomegaly. G-button intact without leakage, redness, or odor.

GU. Pre-pubertal exterior female genitalia. Anus patent and intact sphincter noted. No redness, odor, or diaper rash noted.


Back. Straight and intact. No hair tufts or dimpling noted.

Neuro. All cranial nerves intact. Reflexes are symmetric, 1+ in upper and lower limbs. Sensation appears normal.

Musculoskeletal. Muscle tone, strength, and mass are within normal limits. N.E. sits momentarily (< 2 seconds unattended). She is able to lift her chest up well in the prone position.

Growth/Development. Reaches and grabs for objects and puts them into her mouth. She is unable to crawl when in prone position. Vocal noises were made
but there were no specific sounds or words elicited. No stranger apprehension demonstrated.

Diagnostic Tests

The following test results were reviewed with patient’s mother:

**Initial chromosome study (quick genetic screen).** No abnormalities on the 13, 18, 21, X, or Y-chromosomes.

**Microassay.** 22 paired autosomes and one pair sex chromosomes, XX. No gross chromosomal abnormality detected.

**Fluorescent in Situ Hybridization (FISH):** Two copies of 22q1.11q are present.

**Heterophile Screening:** Normal

Discussion of Findings

A sequence is a group of related anomalies that generally stem from a single initial anomaly that causes an alteration in the surrounding tissues or structures. PRS is identified by micrognathia (small lower jaw) or retrognathia (lower jaw displaced to the back), glossoptosis (displacement of the tongue into the throat), and a U-shaped or cleft palate. During the 7th-11th week of embryonic development the mandible pushes the tongue superiorly, prevents fusion of the palatal shelves, and causes the malformed or absent palate (Shur & Abuelo, 2009). It occurs in 1:8,500 live births, is more frequent in males, and has a greater incidence among twins. PRS can occur as an isolated event, in which heredity is not a factor. Examples of isolated, or non-syndromic causes of PRS are: intrauterine positional malformation, amniotic bands, uterine fibroids, maternal alcohol use, and teratogenic exposures. PRS can also occur due to a mode of inheritance, occurring in association with other syndromes. Examples of syndromic
causes of PRS include Stickler, DiGeorge (velocardiofacial), and Treacher-Collins syndromes (Evans, Rahbar, Rogers, Mulliken & Volk, 2006).

Respiratory distress is the greatest concern with PRS patients. Primary management is determined by the severity of the respiratory manifestations. Some patients can be managed with non-surgical interventions, such as prone positioning and use of a nasopharyngeal tube. In severe cases, surgical intervention is necessary. Some patients may also require a tracheotomy (Cole, Lunch & Siator, 2008). N.E. exhibited a progressive manifestation of respiratory episodes that impaired her ability to breathe and eat. Failure to correct recurrent episodes of hypoxia could result in permanent cognitive deficits (Drescher et al., 2008). Her situation was further compounded by her cleft palate. Cleft palate accompanies PRS in approximately 50% of reported cases (Jones & Smith, 2006). Mandibular distraction osteogenesis is a surgical procedure that allows the jaw to lengthen and provide greater room on the floor of the mouth for the tongue to rest. This removes the airway obstruction by the tongue. This surgical procedure is very effective and once the bones have begun to fuse, the metal appliances are removed (Evans et al., 2006). N.E. is scheduled to have her appliances removed in November 2009.

Many infants with PRS experience weight loss and a high rate of failure to thrive is reported (Evans et al., 2006). After the jaw distraction N.E. was unable to suck on her specialized bottle nipples, so the G-button provided an alternate route for nutritional supplementation. N.E.'s parents have received instruction and support from both the nutritionist and a cleft palate nurse educator, which has enabled her weight to steadily increase. Today, her weight is WNL for her age and height. Besides complicating
feeding, a cleft palate can cause an increase in ear infections and varying degrees of hearing loss. Insertion of PE tubes prevents the recurrence of otitis media and scar tissue on the tympanic membranes and optimizes her hearing ability (Tinanoff, 2007). To date, the tubes appear to be functioning well.

At the first visit with the geneticist (Dr. Kukolich) in September, initial genetic tests were ordered. Approximately 50% of PRS cases are attributed to genetic heritability. The initial chromosome study reveals matching pairs of chromosomes 13, 18, and 21. The patient also has a matching pair of X chromosomes and no Y chromosome. The major trisomy disorders (Trisomy 13, 18, and 21) and chromosomal aneuploidies (Turner, Kleinfelter, 47XXX and 47 XYY) have been associated with cleft palate and PRS (Descartes & Carroll, 2007). N.E. did not have any of these syndromes. The microassay reveals 22 matching pairs of autosomes and one pair of female sex chromosomes. The mother was reassured the results of the microassay were normal. A microassay is not a stand-alone test in the diagnostic lab because it is unable to detect small rearrangements, microdeletions, or low levels of mosaicism (Lewis, 2008). This lead to the third genetic test ordered in order to determine if N.E. carried a deletion in the 22q1.11q. chromosome. This deletion is responsible for GGT1 or GGT2 (OMIM 231950; OMIM 137181), a specific location on the chromosome that identifies protein miscoding in certain cleft palate anomalies. Defects on this chromosome can produce alterations in immune function and can be fatal without proper identification and treatment (Jones & Smith, 2006; Lewis). A FISH test was performed to determine if two complete copies of 22q1.11q were present. Once again, N.E.'s mother was reassured that two complete copies were present and that her results were normal. Unfortunately,
PRS occurs in many other malformation syndromes that are unable to be detected by the previous three tests. If the geneticist suspects other hereditable syndromes, additional FISH testing must be ordered (Jones & Smith).

Assessment

Diagnoses

Acute Diagnosis.

None identified at this visit.

Chronic Diagnoses.

1. Pierre Robin Sequence (765.0). N.E. was born with micrognathia, glossptosis, cleft palate, and airway obstruction. Micrognathia is the initial anomaly that affects the surrounding tissues and structures in the mouth (Cole et al., 2008).

2. Complete cleft palate (749.21). N.E. was born with a cleft palate as a result of her micrognathia and glossoptosis. Cleft palate accompanies heritable as well as non-heritable conditions that must be further investigated by the health care provider (Evans et al., 2006).

3. Developmental Delay (783.40). Monitoring specific milestones are important for determining the presence of developmental delay. These milestones encompass cognitive, fine motor and gross motor skills. N.E.’s inability to crawl or sit demonstrates gross motor delay. Her inability to suck on her bottle nipples or take food from a spoon demonstrate fine motor skill deficit. It is difficult to determine her cognitive level of functioning, so the healthcare
provider must address her progress at each healthcare visit (Feigelman, 2007).

4. Speech Delay (315.39). Although her mother reports that she is “babbling”, N.E. did not demonstrate this in the office. Since language acquisition begins to emerge by six months of age, recognition of red flags in conjunction with barriers to speech (cleft palate, difficulty swallowing) allow the patient obtain early access to intervention, such as speech therapy (Feigelman, 2007).

Differential Diagnosis.

Stickler Syndrome (759.87). Stickler syndrome (Hereditary Arthrophthalmopathy, OMIM 184840) should be considered in any infant presenting with PRS. Stickler syndrome is the most common genetic syndrome that accompanies PRS. Although it is a rare syndrome, affecting 1:7500 newborns, PRS is a comorbid condition that is present in approximately 30% of Stickler syndrome individuals. It is a collagen disorder that affects the ocular, craniofacial, cardiac, and musculoskeletal systems. It is autosomal dominant with incomplete penetration, making the systemic features of Stickler syndrome highly variable and affected and unaffected family members are often indistinguishable. In her pedigree, N.E. has family members with myopia and cataracts. Myopia is present 90% of the time and non-traumatic retinal detachment usually occurs before the age of 10. Thirty-eight percent adults were diagnosed with Stickler syndrome at the time of their first retinal detachment. Seventy percent of these individuals experience hearing loss and forty-six percent have a mitral valve prolapse. Affected patients exhibit flat facies, long fingers, and joint pain that mimic juvenile rheumatoid
arthritis. The geneticist notes that N.E. and her mother have long fingers (Lee & Hayward, 2008; Smith & Jones, 2006).

Impressions

N.E. is gaining weight and has not had any recurrence of respiratory distress. She needs continual work to improve her sucking and chewing abilities. Perhaps this will improve when the jaw distraction appliance is removed. Her oral functioning will need to be reevaluated by the surgeon and the ENT. In some instances, patients with PRS have decreased pharyngeal motor organization and increased dysphagia, requiring some patients to undergo electromyography (EMG) in order to detect muscle dysfunction (Baudon et al., 2009). At this point it is premature to order an EMG, so watchful waiting will determine if it will be needed in the future. In addition to the problems with her oral musculature, careful observation of her teeth eruption is important. Cleft palate can cause teeth eruption to be delayed, malpositioned, or absent. She will require collaboration with a pediatric dentist and orthodontist (Tinanoff, 2007). She also will need to be monitored for her speech development during the next few years. Ruiter, Korstein-Meijer, and Goorhuis-Brouwer (2009) note that children with a cleft palate disorders have greater communication difficulties than children with normally formed palates. They posit that language production and comprehension, hypernasality, and articulation peak during the toddler years.

The patient has a very supportive family who is very attentive to her needs and to her multiple specialist and therapy appointments. I am concerned that she is not enrolled in ECI. This program would benefit N.E., as well as her family, because it provides a large variety of services that N.E. needs at no additional cost. In addition,
these services are performed within the patient’s home. Enrolling N.E. would help alleviate some of the family’s time constraints that her current appointments monopolize. I think that it is also important to consider the needs of N.E.’s family. Many times, the family members of a special needs child tend to ignore their needs because of time and energy constraints. Family planning and genetic counseling N.E.’s parents about the risks for recurrence of PRS and cleft palate will be discussed after her ophthalmologic exam. Based on the current findings, there is a 3-5% risk that PRS and cleft palate will recur. If N.E. is discovered to have Stickler syndrome, there is a 50% chance that future offspring will inherit this condition (Lee & Hayward, 2008; Lewis, 2008).

Plan

Genetics Follow-up

Return to genetics clinic in one year. If abnormal ophthalmologic findings present, the patient will return to the clinic for further genetics testing.

Oral Management

1. Continue to monitor N.E.’s airway. Teething may cause increased salivation. Follow with oral surgeon in November the removal of jaw appliance.

2. Dental hygiene. Begin to brush teeth as they erupt. Follow with pediatric dentist.

3. Continue to use soft flexible nipple of choice when feeding through G-button to promote the use of oral musculature. Continue to encourage soft foods on spoon, offering a variety of textures and flavors. Follow up with pediatric plastic surgeon when patient is 12-months old for cleft palate closure.
4. Follow with neurologist if swallowing and chewing deficits continue after
distraction appliances removed and cleft palate is repaired.

*Rationale:* The patient with PRS and cleft palate can present complicated
management for the DNP. Assuring a continual patent airway is vital to the
prevention of hypoxia and future cognitive deficits. Reinforcing the importance of
daily dental hygiene, limiting sweetened drinks and foods, and attending dental visits
are important components that the DNP can assess for during routine examinations.
Encouraging the parents to continue to offer soft nipples and foods provide the
patient with the opportunity to improve oral musculature and coordination (Tinanoff,
2007).

**Referrals**

1. Pediatric ophthalmology referral.

*Rationale:* There is tendency for providers to under recognize Stickler syndrome.
The ocular manifestations in Stickler syndrome are highly penetrant and an eye
examination is essential in making the diagnosis in some patients. Patients with
Stickler syndrome have a distinctive vitreous appearance and make the eye
examination an important component of diagnosis and directing genetic testing for a

2. Continue regularly scheduled appointments with speech and physical therapy.

3. Coordinate and update N.E.’s changing nutritional needs with nutritionist
(formula, foods, and vitamins).

4. Appointment with other specialists: GI, ENT, audiologist, plastic surgeon, oral
surgeon, and neurologist.
Rationale: Complex teamwork is necessary when treating pediatric patients with complex issues, such as PRS and cleft palate. The interrelatedness of hearing and speech are dependent upon coordination of care within multiple specialty groups. The DNP, when functioning in the role of primary care provider, must help to coordinate multidisciplinary care for the complex pediatric patient (Ruiter et al., 2009).

Lab

1. Additional FISH COL2AI located on chromosome 12q 13.11-13.2 (if ophthalmologic findings are abnormal).

Rationale: Appropriate genetic testing should be conducted on any infant that has PRS, cleft palate, and an abnormal ophthalmologic examination. There should be a high suspicion of Stickler syndrome when patients present with myopia and ocular lesions. Geneticists look for COL2AI, which is located on the 12q13.11-13.2 chromosome. If COL2AI has missing alleles, the diagnosis of Stickler syndrome is confirmed. This test is costly ($2,500-5,000) and there is currently one collagen laboratory in Pennsylvania that processes this test. Prior authorization from Medicaid is necessary before this test can be conducted. If the ophthalmologic exam reveals myopia or early retinal detachment, the provider should make every effort to have this test ordered. Identifying the diagnosis of Stickler syndrome is important for the preservation of vision, supportive care for musculoskeletal deformities, recognition and treatment of hearing loss, and the provision of genetic counseling for families (Lee & Hayward, 2008; Shur & Abuelo, 2009; Smith & Jones, 2006).

Anticipatory Guidance
1. Follow with general pediatric healthcare provider for routine wellness examinations and for signs and symptoms of ear infections

*Rationale: Craniofacial malformations preclude the patient to an increased risk of otitis media and hearing loss. Early detection and management are recommended for optimal speech, hearing, and developmental outcomes (Drescher et al., 2008; Evans et al., 2006).*

2. Receive recommended immunizations at 6-month well exam

3. Seasonal and H1N1 influenza vaccine for patient (at 6-months) and for immediate family

4. Basic growth and development teaching regarding physical, cognitive, and emotional expectations for patient.

5. Family planning

*Rationale: The DNP must appreciate and treat the whole patient, which includes basic treatments and teaching to assure optimal growth and development. As N.E. approaches 6-months, she will be expected to reach certain milestones, including stranger apprehension and independent separation from her mother. Teaching the parents about these expected behaviors allows for smooth transition and healthy outcomes (Feigelman, 2007). Patients with PRS and craniofacial deformities are at a greater risk for cognitive, behavioral, and emotional difficulties. The role of the DNP includes identifying these deficits and helping the family adapt to these findings (Drescher et al., 2008).*

**Family support**

1. Referral to ECI
2. Referral to cleft palate and PRS support groups

Rationale: Through the Texas Department of Assistive and Rehabilitation Services (2007), ECI provides no-cost services for patients enrolled in Medicaid. Programs are available for those with cognitive, motor, communicative, and social-emotional delay. N.E. also has dysphagia and cleft palate, which enable her to receive more in-depth ECI services. The DNP can refer a patient on-line or the family can call for the referral at their convenience. Support groups help to reduce feelings of isolation and despair, which are commonly experienced by parents with special needs children (Drescher et al., 2008; Tinanoff, 2007).

Patient Handouts:

2. ghr.nlm.nih.gov/condition=sticklersyndrome

Rationale: The DNP must provide high quality, reliable information for individual patients and their families. The DNP must consider the reading literacy and health literacy levels so that the information given is understandable. If the family does not have Internet access, the DNP should provide hard copies of the above documents at the office visit (United States Department of Health and Human Services, 2008).
References


Appendix

Legend:
- = Normal female, male
- = Female, male who expresses trait
- = Female, male myopia
- = Female, male cataracts
- = Female, male long fingers
- = Terminated pregnancy