Case Study 1: Trisomy 21 with Congenital Cataract

NURS 6045 Practicum II

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Preliminary Information

Selection of Case

I chose this case because, until now, I had not encountered a congenital cataract in a newborn. The reported prevalence of congenital cataracts in developed countries is rare, with 1 to 3 in 10,000 children (McCreery, 2010). This cataract was found while performing a routine newborn eye assessment with an ophthalmoscope, looking for the red reflex. I wanted to share this information with my peers, in the event that an infant or child with undiagnosed cataracts is seen in their clinic.

Cataracts are one of the leading causes of blindness in children. Early detection and prompt intervention, especially in newborns, is critical for optimal visual outcome. Prompt referral to an ophthalmologist is essential if a cataract is suspected (McCreery, 2010).

Cataracts may not be present at birth, but can develop over time, in children and may be associated with a number of systemic and ocular disorders (McCreery, 2010).

Type and Number of Encounters

This infant delivered in a small community hospital. The first encounter was at delivery. Infant was admitted to the Neonatal Intensive Care Unit (NICU) secondary to respiratory distress, requiring oxygen by nasal cannula, dysmorphic features, and cataract in his left eye. Infant was discharged and scheduled for readmission in two weeks when the eye surgeon planned to perform cataract surgery.

Baby K.W. was readmitted to the NICU on day of life 13 for cataract surgery the next morning.
Insurance

This infant was covered by Tricare Insurance.

Subjective

Patient Profile

This newborn male infant delivered by spontaneous vaginal delivery.

Background Information

CC. K.W. is a newborn Hispanic male, who delivered at 39 2/7 weeks gestation, with meconium stained amniotic fluid. He was admitted to the NICU from the delivery suite for respiratory distress and rule out sepsis.

HPI. This infant was delivered by spontaneous vaginal delivery.

PMH.

Birth. K.W. was 39 2/7 weeks gestation, delivered by spontaneous vaginal delivery to a gravida 2, Para 1, 25 year old Hispanic female, who received prenatal care at the Prenatal Clinic. The mother had a negative Quad screen. Prenatal ultrasound showed an echogenic focus in the left heart ventricle which was thought to be a normal variant, when coupled with the negative Quad screen. There was no exposure to medications, alcohol, recreational drugs, or tobacco. Her prenatal screening labs were negative.

Birth weight: 3.48 kg (7 lbs. 6.6 oz.)

Length: 50.5 cm (20 in.)

Head circumference: 33.5 cm. (13.25 in.)

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

K.W. delivered by vaginally, with meconium stained amniotic fluid. He cried spontaneously at the perineum, good heart rate and tone, fair respiratory effort with mild
nasal flaring. Color was pink with bulb suctioning, drying, and stimulation. He was noted to have dysmorphic features concerning for Trisomy 21, to include: upslanting palpebral fissures, low set ears, simian creases and clinodactyly of the fifth digits of both hands.

He was transferred to the NICU for transition and further evaluation.

Medications

K.W. received Erythromycin ophthalmic to both eyes at delivery. He also received Vitamin K, IM, at delivery.

Family History.

Mother. Hispanic female who speaks English. She does not work outside the home. She reports no health problems.

Father. Caucasian male who speaks English. He is active duty in the military. He reports no health problems.

Additional family information

There is no known family history for Down syndrome (Trisomy 21). There are no other known anomalies. Mother’s quad screening was negative for open neural tube defect, Down syndrome and Trisomy 18.

Social History

Parents are married, with a 3 year old female child living in the house. Infant will be discharged home with parents following hospital discharge. Parents deny use of tobacco or recreational drugs. Both maternal and paternal grandparents live in another state. Social worker is involved for family support. Parents were unaware that this infant was
at risk for Trisomy 21. Mother desires to breastfeed infant, but is not opposed to formula if needed.

Review of Systems on Admission.

**Head/Neck:** Normal appearing head, with slight molding. Thick nuchal fold present. Anterior fontanel is open, soft and flat. Eyes with upturned palpebral fissures. Red reflex present in the right eye, unable to elicit in the left eye. Nose normally placed, nares patent, with mild flaring. Nasal cannula in place. Palate intact, slightly protuberant tongue. Ears low set, no pits or tags noted.

**Skin:** No rashes, bruising or lesions noted.

**Cardiac:** Regular rate and rhythm. No murmurs. S1S2 noted. Femoral and posterior tibial pulses 2+ and equal bilaterally.


**Abdomen:** Soft, flat, with no masses or organomegaly. Three vessel cord noted.

**GU:** Normal male genitalia. Testes descended bilaterally. Anus is patent.

**Neuro:** K. W. has normal activity for gestational age. Tone is hypotonic. She has normal reflexes.

**Extremities:** Moves all extremities. Clinodactyly of 5th digits of hands bilaterally. Single palmar crease bilaterally. Increased space between the first and second toes bilaterally.

**Growth/Development:** Infants birth weight is average for gestational age per infant growth chart.
Objective

Physical Examination

**General:** This is an active, alert, male infant in a radiant warmer, on nasal cannula at 2L/min at 30%.


Blood pressure: 67/44.

**Weight:** 3.480 kg (7 lbs., 6.6 oz.).

**Length:** 50.5 cm (20 in.).

**Head Circumference:** 33.5 cm. (13.25 in).

**Head/Neck:** Normal appearing head. Anterior fontanel is open, soft and flat. Neck has thick fold. Jaw appears normal.

**Skin:** No rashes, bruising or lesions noted.

**Eyes:** Eyes with upslanting palpebral fissures bilaterally. Pupils equal and react to light. Red reflex is present in right eye, but abnormal in the left eye.

**Ears:** Ears low placed. No pits or tags noted.

**Nose:** Nose normally placed. Nares are patent. Nasal cannula in place.

**Oropharynx:** Palate intact. Slightly protruding tongue. No abnormalities noted in mouth or throat.

**Cardiac:** Regular rate and rhythm. Normal S1S2. No murmurs noted. No palmar pulses noted. Good femoral and posterior tibial pulses. Infant is well perfused.

**Lungs:** Chest is symmetric. Nipples are normally placed. Breath sounds are equal and course, with good air movement noted. Infant’s respirations are slightly labored on 2 L/min nasal cannula set at 30%.
GI: Soft, flat, with no masses or organomegaly. Bowel sounds are present. Three vessel cord is present. Nipple fed adequate volume without difficulty.

GU: Normal male genetalia. Anus is patent. Infant has voided and stooled since birth.

Extremities: Infant moves all extremities well. Clinodactyly noted of 5th digits of both hands. Single palmar crease noted bilaterally. Increased space between first and second toes bilaterally. No hip clicks or clunks noted.

Back: Spine is straight and appears normal. No dimples noted.

Neuro: This infant has normal activity for gestational age. Good suck and gag present. Normal reflexes elicited.

Musculoskeletal: Hypotonic.

Diagnostic Tests.

Complete blood count (CBC): K.W. has normal hemoglobin and hematocrit. No thrombocytopenia or polycythemia noted. CBC was not a significant indication of sepsis.

Up to 66% of newborns with DS have thrombocytopenia and up to 33% have polycythemia. Children with DS have an increased risk to develop leukemias

(Weijerman & deWinter, 2010).

Chromosomes: This infant had the following findings consistent with Trisomy 21:

Upslanting palpebral fissures            Thick nuchal fold
Low set ears                             Single Simian creases bilaterally
Increased space between first and second toes bilaterally
Clinodactyly of the fifth digits of both hands

The characteristics and clinical signs at birth of Trisomy 21 can guide the decision to perform chromosome testing for the confirmation of a Down syndrome
diagnosis. Neonatal signs of Trisomy 21 include: small ears, wide space between 1st and 2nd toe (“sandal gap”), nuchal skin fold, upward slant of the eye split, and simian crease (Weijerman & deWinter, 2010; Jones, 2006).

**Echocardiogram:** K.W.’s echocardiogram showed no cardiac abnormalities despite a prenatal ultrasound that showed an area of echogenic focus in the left ventricle.

*All newborns with Down syndrome (DS) should be evaluated for congenital heart disease in consultation with a pediatric cardiologist. An echocardiogram is recommended to detect abnormalities that may not be symptomatic or apparent on physical examination”* (Roizen & Stark, 2010). The prevalence of congenital heart defects in neonates with DS is about 44-58% (Weijerman & deWinter, 2010).

**Eye Exam:** K.W. had an abnormal red reflex in the left eye. A cataract was confirmed by the ophthalmologist.

*More than half of children with Trisomy 21 have ocular abnormalities.*

*Congenital cataracts comprise 4-7% of these disorders. An early start to visual screening is essential for detecting defects, and especially important in finding congenital cataracts (Weijerman & deWinter, 2010).*

**Hearing Screen:** K. W. passed his neonatal hearing screen, but will need serial follow-up examinations with the audiologist.

*Hearing impairment is prevalent in children with DS. Even mild hearing loss can influence developments and affect speech. Regular assessment of hearing function is very important (Weijerman & deWinter, 2010).*
Newborn Screen: K.W.’s first and second newborn screens were normal.

*Thyroid disorders have been reported in up to 28-40% of children with DS.*

*These disorders increase in frequency (up to 54%) with age. Hyperthyroidism (Grave’s Disease) also occurs in children with DS (0-2%)* (Weijerman & deWinter, 2010).

**Discussion of Findings**

Down syndrome (DS) is a congenital abnormality caused by trisomy of the genes on chromosome 21 (Creavin & Brown, 2008). Down syndrome is the most common chromosome abnormality among live births (8%). It is also the most common form of intellectual disability (Roizen & Stark, 2010; Weijerman & deWinter, 2010). Because of medical advances, the survival of persons with DS has increased. This longer life expectancy requires caring for these persons over their longer lifespan.

Down syndrome is characterized by a number of dysmorphic features and delayed psychomotor developmental delays. Children with DS also have an increased risk of congenital defects and organic defects including congenital heart defects, gastrointestinal defects, celiac disease and thyroid abnormalities. Congenital heart defects and respiratory infections are the most frequently reported medical disorders responsible for death in persons with DS. The average age at death of persons with DS in the United States rose from 25 to 49 years between 1983 and 1997 (Weijerman & deWinter, 2010).

Medical care of infants with DS is dependent on the specific issues that occur in the individual patient. The prevalence of congenital heart defects (CHD) in babies with DS is 44-58% worldwide. The most common forms of CHD are atrioventricular septal defect (ASD) and ventricular septal defect (VSD). Because of the high incidence of CHD in infants with DS, early
recognition is essential. Early diagnosis leads to optimal defect management and can prevent the
development of pulmonary hypertension. An echocardiogram should always be performed in the
first month of life (Weijerman & deWinter, 2010).

Hearing impairment and otologic problems are common in children with DS. Children
with DS have delayed speech development. In approximately half of children with DS, sleep-
disordered breathing is seen.

Respiratory problems are responsible for the majority of hospital admissions and death in
children with DS. Respiratory syncytial virus (RSV) is seen more frequently in DS children and
puts them at greater risk for hospitalization. Other causal factors for respiratory problems are
airway anomalies (Weijerman & deWinter, 2010).

Gastrointestinal tract disorders are present in 4-10% of children with DS and have an
impact on morbidity during the first year of life. Duodenal atresia and celiac disease are
common defects in babies with DS. Constipation, secondary to hypotonia is a serious problem
with DS children (Weijerman & deWinter, 2010).

Children with DS have a high frequency of various ocular abnormalities. “A cataract is
opacity of the lens of the eye that causes partial or total blindness” (McCreery, 2010). The
occurrence of early cataract among children aged up to 17 years with DS has been reported from
5-50% (Haargaard & Fledelius, 2006). According to several studies of infantile cataracts,
trisomy 21 accounts for a third of the cases (Lim, Rubab, Chan, & Levin, 2010). Cataracts are
one of the leading causes of blindness in children. Early detection and prompt intervention,
especially in newborns, is critical for optimal visual outcome. Prompt referral to an
ophthalmologist is essential if a cataract is suspected. Cataracts may not be present at birth, but
can develop over time, in children and may be associated with a number of systemic and ocular
disorders (McCreery, 2010). Good vision is very important to the development of a challenged DS child. Visual screening is necessary to detect defects that can be treated (Weijerman & deWinter, 2010). Infants with cataracts should undergo surgery in the first 4-6 weeks of life. Children with unilateral cataracts are at increased risk of amblyopia and must adhere to the post-operative schedule of occlusion therapy to achieve a good visual outcome (McCreery, 2010).

Assessment

Diagnoses

Acute Diagnoses

Trisomy 21 (758.0). K. W. was born with multiple clinical characteristics of Trisomy 21 or Down syndrome, to include small low-set ears, wide space between 1st and 2nd toe (“sandal gap”), nuchal skin fold, upward slant of the eye split, and simian crease He also had clinodactyly of the 5th fingers bilaterally. Of infants with clinodactyly, 80% of them have Down syndrome (Weijerman & deWinter, 2010).

Congenital Cataract (743.30). K. W. was born with a cataract on his left eye, requiring surgical removal. According to several studies of infantile cataracts, trisomy 21 accounts for a third of the cases (Lim, Rubab, Chan, & Levin, 2010).

Chronic Diagnoses

Amblyopia (368.0). K. W. is at risk for amblyopia following cataract extraction surgery and placement of aphakic contact lenses. Patching will be necessary to treat amblyopia. Children with unilateral cataracts are at increased risk of dense deprivation amblyopia (McCreery, 2010).

Aphakic Glaucoma (365.14). K. W. is at increased risk for aphakic glaucoma following cataract surgery with placement of aphakic contact lenses (McCreery, 2010). Glaucoma
is a common complication following surgery for cataracts in pediatric patients (Lloyd, Ashworth, Biswas, & Abadi, 2007).

Strabismus (378.60). K.W. is at risk for strabismus following cataract extraction surgery. Studies have reported rates of strabismus as high as 46% following cataract surgery, with the overall incidence in patients with unilateral cataracts being 59%. Findings show a statistically significantly higher incidence of strabismus in unilateral cataract cases compared to bilateral cases (Lim et al., 2010).

**Differential Diagnosis**

There are no differential diagnoses for Trisomy 21 as this diagnosis was confirmed by karyotype testing.

Differential Diagnosis of cataract includes other conditions that cause leukocoria (McCreery, 2010).

1. Retinoblastoma (190.5):

“Retinoblastoma is the most common intraocular tumor of childhood. The majority of cases are diagnosed in children younger than two years of age, and approximately 95 percent before the age of five. Children with retinoblastoma frequently (but not always) present with leukocoria” (Kaufman & Teed, 2010). About 50% of cases are diagnosed by leukokoria.

**Impressions**

K.W. was born with multiple clinical characteristics of Down syndrome (Trisomy 21). He showed no congenital heart defects by echocardiogram. He did not appear to have any gastrointestinal defects and was breastfeeding well without problems.
His newborn screens were normal but will need to be followed for endocrine issues. K.W. is at increased risk for thyroid abnormalities, as well as diabetes. K. W. will need to be tracked for growth pattern and Down syndrome-specific growth curves. He needs special attention weight, to avoid obesity (Weijerman & deWinter, 2010).

K.W will need follow-up with orthopedics. Due to hypotonia and ligamentous laxity, joint and ambulation problems can develop. Acquired hip dislocation occurs in up to one-third of children with DS (Weijerman & deWinter, 2010).

K. W. required cataract extraction surgery at 2 weeks of age. He will need close post-surgical follow-up and care. “Cataract surgery in children requires a firm commitment on the part of the child’s parents to become involved in the post-operative regimen, which can be burdensome. Lack of adherence to the regimen could result in permanent adverse sequelae” (McCreery, 2010).

K.W. should be assessed for the need for the Early Childhood Intervention Program. In the event that developmental or physical delays are assessed, this program would benefit K.W. by providing occupational, physical, speech therapy and a number of other services.

This baby has a very supportive family. Although they were unaware of the situation, they are very attentive to his needs. They have verbalized the importance of long-term follow-up for their son’s health issues.

**Plan**

1. Surgical extraction of cataract from left eye, by ophthalmologist (CPT 66983).

   “Infants with cataracts should undergo surgery as soon as possible, usually in the first four to six weeks to months of life. The visual axis must be cleared by 16 weeks of age to achieve visual acuity of 20/40 or better” (McCreery, 2010).
2. Social service and caregiver support. These parents were unaware of the DS diagnosis prior delivery and are adjusting to the new knowledge. Encourage bonding process by involving parents in infant care.

“The parents or caretakers of a child for whom a diagnosis of DS is being considered or has been confirmed should be informed in a supportive, positive, caring and honest manner. The timing of the disclosure of specific DS-related problems must be balanced with respect for the opportunity for parents to welcome their child” (Weijerman & deWinter, 2010).

3. Evaluate and encourage breastfeeding to assure adequate intake for growth.

“Breastfeeding should be promoted not only because of the psycho-emotional or immunity benefits but particularly because breastfeeding has specific advantages for children with DS in terms of stimulating the development of the oral motor system. However, because of their impaired oral motor function, children with DS can have problems with drinking, swallowing, and chewing” (Weijerman & deWinter, 2010).

Continuity of Care and Referrals

1. Post-operative follow-up and care for cataract surgery to include application of eye drops as ordered by the ophthalmologist, frequent office visits and long-term occlusion therapy for amblyopia. K.W. needs to be closely monitored success of the surgery and for complications such as infection and associated ocular anomalies.

“Cataract surgery in children requires a firm commitment on the part of the child’s parents to become involved in post-operative care. Adherence to the post-operative schedule of occlusion therapy is essential to achieving good visual outcome” (Weijerman & deWinter, 2010).

*Measurements should be plotted on the appropriate special growth chart for children with DS. “This will enable detection of additional disturbances of growth associated with other disorders, such as hypothyroidism or celiac disease, and the monitoring of excess weight gain” (Roizen & Stark, 2010).*

3. Continued cardiac evaluation.

*“Continued clinical cardiac evaluation is needed because of the high risk of mitral valve prolapse and aortic regurgitation in adolescents and young adults” (Roizen & Stark, 2010).*

4. Monitor for sleep disorders.

*Up to 75 percent of children with DS have sleep apnea (Drutz, 2010).*

5. Genetic counseling

*Genetic counseling would be beneficial for family planning. Needs a genome.*

6. Orthopedic consult for potential joint and ambulation problems

*“Acquired hip dislocation occurs in up to 30% of children with DS and needs special attention. Most of these disorders manifest themselves once children with DS start walking” (Weijerman & deWinter, 2010).*

7. Endocrine consult and long-term follow-up.

*“Thyroid disorders have been reported in up to 28-40% of children with DS and they increase in frequency, up to 54% as the children age. Interestingly, diabetes mellitus develops more frequently (1%) in children with DS” (Weijerman & deWinter, 2010).*
8. Audiology Consult

“Hearing impairment and otologic problems are prevalent in children with DS, and these problems correlate substantially with developmental problems” (Weijerman & deWinter, 2010).

Rationale: When functioning in the role of care provider, the DNP must assist in coordinating multidisciplinary care of the complex pediatric patient.

9. Referral to Early Childhood Intervention as warranted

Rationale: ECI is a service provided through the Texas Department of Assistive and Rehabilitation Services (2007). Programs are available for patients with cognitive, motor, communicative, and social-emotional difficulties. K.W. is at risk for developmental, motor, and functional disabilities.

Anticipatory Guidance

1. Educate family for expectations and complications of Down syndrome

Down syndrome can cause a number of medical complications, but many of them can be treated (Drutz, 2010).

Rationale: The Doctor of Nursing Practice (DNP) must provide high quality, reliable information for patients and families (United States Department of Health and Human Services, 2008).

2. Educate family for importance of follow-up routine care and immunizations.

3. Encourage parents to seek prompt medical care for respiratory illness

Individuals with DS are at increased risk for hospitalization, intubation, and death with the flu or RSV (Perez-Padilla et al., 2010).
Family Support

National Library of Medicine

www.nlm.nih.gov/medlineplus/downsyndrome.html (available in Spanish)

Association for Children with Down Syndrome

www.acds.org

National Association for Down Syndrome

www.nads.org

Rationale: The Doctor of Nursing Practice (DNP) must provide high quality, reliable information for patients and families. The DNP must take into consideration the reading literacy and health literacy levels in order to offer understandable information to patients. The DNP should provide hard copies of documents if the family does not have access to the internet (United States Department of Health and Human Services, 2008).
References


