Intellectual and Developmental Disorders NCLEX Review

I. INTELLECTUAL DISABILITY

Although intellectual disability (ID), formerly called mental retardation, is oft en thought of as strictly a cognitive disability, the definition of the concept has taken on a broader context in recent years. Rather than referring strictly to a child's thought-based abilities, experts refer also to the child's behavioral abilities. As a result, when a person with limited intelligence is able to function relatively normally, he or she should be seen in a different light from someone who has the same intelligence quotient (IQ) but who has a great deal of trouble functioning in society.

Incidence: Th e IQ of 1% to 3% of the population in the United States falls below 70, but up to 85% of that group is shown to have only a mild disability.

Etiology:

Both genetic and environmental causes.

1. Environmental causes.

a. Fetal alcohol syndrome is the number one preventable cause of ID in the United States.

b. Lead exposure: may occur either prenatally and/or as a childhood exposure.

c. Infectious diseases: may occur either prenatally and/or as a childhood exposure.

d. Poor or abusive parenting (e.g., shaken baby syndrome).

e. Perinatal hypoxia that occurs during pregnancy, labor, and/or delivery.

f. Hypoxia of a child that may occur postdelivery, most commonly in premature infants, or as a result of an accident (e.g., near drowning).

2. Genetic causes.

a. Fragile X syndrome.

- Most common genetic cause of ID.
- ii. X-linked recessive syndrome . (1) A Punnett square with an example of the inheritance pattern for fragile X syndrome is shown below. Th e mother is heterozygous for the disease (i.e., she carries one affected X chromosome ["x"X]), and the father is unaffected (XY).

(a) If the off spring is female, there is a 50% probability of carrying an affected X and potential for

exhibiting symptoms of the fragile X syndrome and a 50% probability of having a normal genotype. (b) If the off spring is male, there is a 50% probability of having fragile X syndrome and a 50% probability of having a normal genotype.

b. Down syndrome. i. Trisomy 21 is the most common Down syndrome genotype.

Pathophysiology:

Damage to the cognitive centers of the cerebrum of the brain that has occurred from one of many possible insults, including hypoxic injury, teratogenic insult, or genetic injury.

Diagnosis:

- Prenatal screenings.

1. May detect a fetus that is at high risk of a genetic syndrome.
2. If the screening is positive, diagnostic tests (i.e., chorionic villus sampling or amniocentesis) may be performed.

- Genetic diagnostic tests provide accurate diagnoses of genetic disorders.
- Growth and development screenings (e.g., DDST, Ages and Stages) are performed during early childhood. a. When a child fails to achieve expected milestones, health-care practitioners should refer the child for additional, more sophisticated cognitive diagnostic testing.
- Cognitive diagnosis tests include the StanfordBinet Intelligence Scale (SB5), the Wechsler Preschool and Primary Scale of Intelligence (WPPSI-III), and the Wechsler Intelligence Scale for Children (WISC-III).

1. SB5: for assessing age 2 through adulthood. i. Includes a comprehensive assessment of intelligence of the child.
2. WPPSI-III: for assessing children 2 years 6 months to 7 years 3 months of age.
3. Includes a number of subscales to provide a comprehensive assessment of intelligence of the young child.

- WISC-III: for assessing children over the age of 6. i. Includes 13 subscales for comprehensive assessment of intelligence.
- Tests for children under 2 are less predictive.
- Some children are not diagnosed until in school when they have difficulty in academic achievement.
- f. Signs and symptoms.

i. To be identified as intellectually disabled, children must have exhibited cognitive impairment, with an IQ below 70, before the age of 18.

ii. Because of the multifactorial focus in relation to ID in recent years, to be labeled as having an intellectual deficit, a child not only must have a below 70 IQ but also must exhibit deficits in “adaptive behavior as expressed in conceptual, social, and practical adaptive skills” (AAIDD, 2013). In other words, the child must have difficulty in other aspects of his or her life (e.g., communicating with others, performing self-care skills, performing employable skills).

iii. Comorbidities are commonly seen in children with cognitive deficits, including sensory deficits, seizure disorders, behavioral problems, and psychological disorders.

iv. Because they are so vulnerable, children with cognitive defi cits are at high risk for physical, emotional, and/or sexual abuse.

Treatment:

1. Repeated growth and development screenings.
2. Early intervention, especially educational stimulation programming, is key but is oft en dependent on the accessibility of resources.
3. Children with ID must be assessed for comorbidities, and, if they exist, they must also be treated (e.g., hearing aids, glasses).

Nursing Interventions:

1. Deficient Knowledge.

- Preconception counseling is essential.

i. Educate clients to avoid exposure to lead and alcohol before getting pregnant and throughout the pregnancy.

- Educate clients regarding lead poisoning prevention strategies for children after delivery.
- Prevent injury from shaken baby syndrome through education programs.
- Prevent injury from drowning by educating parents regarding the need for early child swim instruction.
- Refer couples who are at high risk for delivering a baby with a genetic defect for genetic counseling.
  2. Risk for Altered Parenting/Grieving.
- Allow parents to express their grief, anger, and/or frustration regarding caring for a child with ID.
- Carefully assess parenting behaviors.

i. Children with cognitive deficits are at very high risk for abuse and neglect.

- c. Refer the family to supportive organizations (e.g., National Down Syndrome Society, National Fragile X Foundation, American Association of Intellectual and Developmental Disabilities).

3. Many additional nursing diagnoses related to the cognitive deficit may be appropriate including Delayed Growth and Development, Deficient Knowledge, Impaired Coping, Self-Care Deficit, Impaired Memory, Risk for Injury, Risk for Self-Mutilation, and Impaired Verbal Communication.

- It is essential to assess growth and development, especially growth and development milestones, to determine the extent of the child’s disability.
- It is very important to relate to the child at his or her functional level rather than the child’s chronological age.
- Refer the child to programs that provide early educational intervention.

- Depending on additional deficits exhibited by the child, refer the family for specialized care, e.g., occupational therapy, physical therapy.
- Provide children with clear, simple explanations of all tasks/treatments.

II. DOWN SYNDROME

Incidence:

1. Th e risk of birthing a child with Down syndrome increases with maternal age. Probability:

- At age 25: 1/2,500 births.
- At age 30: 1/1,000.
- At age 40: 1/100.
- At age 49: 1/10.

Etiology:

1. The most common cause of Down syndrome is the nondisjunction of chromosome 21 during meiosis.

- Three number 21 chromosomes, called trisomy 21, end up in the nucleus of the zygote and in the growing embryo and fetus
- Th e child, therefore, has a total of 47 chromosomes in the cells of his or her body.

2. Down syndrome may also occur as a result of a chromosomal translocation, including chromosome 21.

Pathophysiology:
1. Because of an excess of chromosome 21 genetic material, the characteristic features of Down appear as:
a. Cognitive deficits.

b. Facial and cranial deformities.

- Slanted eyes.
- Wide, flat nasal bridge.
- Protruding tongue.
- Small, low-set ears.

c. Muscular hypotonia, poor muscle tone throughout the body, often resulting in feeding difficulties, recurrent respiratory illnesses, obesity, and protruding abdomen.

d . Simian creases: unbroken “life lines” that stretch across the palm of the hand.

e. Lax joints, often resulting in joint injuries.

f. Also at high risk for cardiac and other congenital defects, including of the gastrointestinal and central nervous systems; Leukemia: 15 times the incidence of the general population; Early onset Alzheimer's disease.

Diagnosis:

1. Prenatal.

- Prenatal screening provides fairly accurate probability of carrying a child with Down syndrome.
- Diagnostic testing is definitive.
- Genetic analysis: either via chorionic villus sampling (CVS) or amniocentesis.

2. Neonatal.

- Clinical picture is suggestive.
- Genetic analysis is diagnostic.

Treatment:

1. Surgery to correct any congenital defects.
2. Repeated growth and development screenings.
3. Early intervention to promote learning and optimal social and behavioral skills.
4. Genetic counseling to provide the couple with information regarding the probability of conceiving a Down syndrome baby in the future.

Nursing Interventions:
1. Deficient Knowledge/Risk for Caregiver Role Strain.

- Educate the parents regarding the genetic etiology of disease.
- Refer the parents for genetic counseling.
- Introduce the parents to another family with a Down syndrome child.
- Provide a referral to an appropriate organization (e.g., National Association for Down syndrome).

2. Risk for Imbalanced Nutrition: Less than Body Requirements (infancy)/More than Body Requirements (childhood).

- Educate the parents regarding the child's poor muscle tone.
- During infancy:

i. Educate the parents regarding the need to feed the baby slowly (if bottle-fed) or refer the breastfeeding mother to an IBCLC (International Board Certified Lactation Consultant) for assistance with latch and milk transfer.

ii. If bottle-fed, the child may need specialized feeding devices (e.g., Haberman feeder) to facilitate feeding.

iii. Refer the parents to an occupational therapist, if needed.

- As the child grows, to prevent obesity:

i. Educate the parents to feed the child a diet with a minimal number of empty calories.

ii. Educate the parents to encourage the child to engage in a daily exercise routine.

3. Risk for Altered Gas Exchange/Ineffective Airway Clearance.

- Educate the parents to seek medical care whenever the child develops an upper respiratory infection (URI).
- Educate the parents to perform respiratory PT to prevent URIs and pneumonia.

4. Risk for Injury.

- Refer the parents to a specialist to determine the potential for neck and joint injuries.
- Encourage the parents to have the child participate in safe physical activities in order to maximize muscle tone and joint health.

III. FRAGILE X

Incidence:

1. Fragile X syndrome is the most common genetic form of ID.
2. Most commonly seen in boys, but girls do exhibit some characteristics of the syndrome.

Etiology:

1. X-linked recessive disease.

- Most severe form seen in males.
- Females are carriers of the syndrome and do exhibit some characteristics of the syndrome.

Pathophysiology:

1. Physical defects are often overlooked.

- Long, narrow face.
- Large ears.
- Lowered epicanthal folds.
- Also may have enlarged testes, lax joints, and mitral valve prolapse.

2. Cognitive Defect.

- Males: moderate to severe deficits.
- Females: because they possess one normal X chromosome, females usually only exhibit mild to moderate cognitive deficits.

3. Behavioral characteristics.

- One-third of children with fragile X will exhibit behaviors related to autism spectrum disorders (see the “Autism Spectrum Disorders” section).
- Aggression.
- Agitation.

Diagnosis:

Diagnosis is often missed, especially in girls.

1. Physical appearance and behavioral characteristics are suggestive.
2. Genetic testing is diagnostic.

Treatment:

1. Repeated growth and development screenings.
2. Early intervention to promote learning and optimal social and behavioral skills.
3. Genetic counseling is essential for any child exhibiting symptoms of autism spectrum disorders and, if a diagnosis is made, for the parents in order to plan for future pregnancies.

Nursing Interventions:

- Deficient Knowledge.

1. Educate the parents regarding the genetic etiology of the syndrome.
2. Refer the parents for genetic counseling.

- Caregiver Role Strain/Risk for Dysfunctional Family Processes/Risk for Injury/Risk for Self-Mutilation.

1. Assess impact of the adverse behaviors on the family.
2. Provide a referral to a facility where expert care/education is provided.
3. Introduce the parents to another family with a child with fragile X syndrome.
4. Provide a referral to an appropriate organization (e.g., National Fragile X Foundation, American Autism Association, American Autism Society).

IV. FETAL ALCOHOL SYNDROME

Fetal alcohol spectrum disorders (FASD) are divided into three subcategories:

Name	Characteristics
Fetal Alcohol Syndrome	Most severe form of FASD, characterized by a wide range of signs and symptoms, including physical, intellectual, and behavioral problems.
Alcohol-related neurodevelopmental disorder	Cognitive and behavioral signs and symptoms
Alcohol-related birth defects	Physical Alterations

Incidence:

Known incidence is approximately 1/1,000 live births, but the incidence is believed to be much higher.

Etiology:
1. Alcohol intake during pregnancy.
2. There is no known safe level of alcohol intake during pregnancy

Pathophysiology:

1. May occur with daily alcohol consumption or with binge drinking.
2. Th ere are a myriad of physiological and psychological signs and symptoms associated with FASD. a. Physiological.

i. Head and facial anomalies .

- Smooth philtrum.
- Microcephaly.
- Short palpebral fi ssures.
- Hypoplastic upper lip.

ii. Small for gestational age.

iii. Organ defects, including:

- Cardiac, especially septal, defects.
- Vertebral malformations.
- Cleft lip and/or palate.
- Renal anomalies.
- Short fingers.
- Sensory deficits.

b. Psychological and behavioral.

i. Low IQ.

ii. Hyperactivity.

iii. Learning disabilities.

iv. Poor reasoning abilities.

Diagnosis:

Absence of a genetic defect that would explain the disorder. Evidence of alcohol consumption during pregnancy, either by self-report, third-party report, and/or toxicology report in combination with clinical evidence

Treatment:

1. Prevention.

- Preconception counseling regarding the need to abstain from any alcohol while trying to become pregnant until the birth of the baby.

2. Substance abuse counseling for women of childbearing age regarding the need to change behavior:

- To prevent FASD. b. In order to provide optimal parenting of the FASD child.

3. Treatment of the injured child.

- Surgery to correct any congenital defects.
- Repeated growth and development screenings.
- Early intervention to promote learning and optimal social and behavioral skills.

Nursing Interventions:

1. Deficient Knowledge.

- Provide preconception counseling regarding the importance of avoiding all alcohol from the cessation of use of birth control until the birth of the baby.

2. Impaired Growth and Development.

- At each well-child visit, it is essential to assess the child’s growth and development.
- Report any deviations from normal to the primary health-care provider.
- Refer the family for expert intervention, as needed.

3. Impaired Social Interaction/Impaired Verbal Communication, especially important during hospitalizations.

- Have the same nurse care for the child as much as possible.
- Establish a routine that is as close to the child’s normal as possible.
- Use alternate means of communication (e.g., pictures) as a way to interact with the child.
- Strongly encourage a family member to accompany the child at all times.

4. Risk for Injury.

- Maintain as safe an environment as possible.
- Provide the child with constant supervision.
- Provide the child with safety equipment when needed (e.g., helmet for head banging).

V. NONORGANIC FAILURE TO THRIVE

Failure to thrive (FTT) refers to a child who is growing and developing much slower than would be expected. Th e child's growth pattern is well below the expected curve on growth charts, and his or her developmental growth is delayed. In many cases, a physical (i.e., organic) reason is diagnosed to explain the poor growth pattern. When no physiological abnormality is present, the diagnosis of nonorganic FTT (NOFTT) is made.

Incidence:

Some experts report that as many as 10 out of every 100 children exhibit FTT.

Etiology:

- Usually a combination of factors. Some experts believe that NOFTT may have both a physiological and a behavioral origin.
- Less than optimal care provided by the baby's primary caregiver, characterized by unresponsiveness to the baby's feeding cues and needs and/or failure to provide stimulation or opportunities to achieve normal behavioral milestones, are factors in the problem's etiology.

a. Behaviors oft en exhibited by the caregiver include:

1. Depression.
2. Substance abuse.
3. Lack of knowledge regarding childrearing skills.
4. Lack of resources.
5. Poor bonding.

Pathophysiology:

- Physiological indicators.

1. Below the 5th percentile for height and/or weight on growth charts or a marked drop in physiological growth.
2. Failure to achieve standard developmental milestones.
3. Poor muscle tone.
4. Abdominal distension.
5. Signs of malnutrition.

- Behavioral indicators.

1. Poor eye contact.
2. Failure to seek parental consolation.
3. Failure to exhibit age-appropriate fear of strangers. d. Disinterest in environmental stimuli. e. Autistic-like behavior.

Diagnosis:

1. Clinical signs and symptoms with no organic reason for the findings.
2. Responsiveness to intervention a. When health-care providers exhibit appropriate parenting behaviors, modeling them for the parents.

Treatment:

1. Provide parenting classes to primary caregivers.

- Parenting classes can be both a prevention and a treatment strategy.

2. Improved nutrition: Affected infants are usually fed high-calorie formula (24 kcal/oz rather than 20 kcal/oz).

3. Multivitamin supplements.

Nursing Interventions:

1. Deficient Knowledge/Impaired Parenting/ Ineffective Role Performance.

- Conduct a thorough psychosocial assessment to identify high-risk families.
- Refer primary caregivers for needed services, optimally before the baby is born, (e.g., substance abuse counseling, fi nancial support counseling, psychological intervention).
- Carefully assess parenting behaviors of primary caregivers.
- Provide parenting education prenatally and/or postpartum, as needed.
- Role model appropriate parenting behaviors during all nurse-parent interactions.

2. Impaired Growth and Development.

- At each well-child visit, assess the child's growth and development.
- Report any deviations from normal to the primary health-care provider.
- Refer the family for expert intervention, as needed.

3. Imbalanced Nutrition: Less than Body Requirements.

- Educate the parents about the nutritional needs of the baby, especially if the mother is breastfeeding.
- Provide a high-calorie formula at each feeding, if needed.
- To prevent unnecessary distractions, educate the parents to feed the baby in a low-stimulation environment.

VI. AUTISM SPECTRUM DISORDERS

Incidence:

- Estimates range widely. The highest incidence estimates of autism spectrum disorders

(ASD) are 1 in 88 to 1 in 150 children.
- Four to five times more likely in boys than in girls (many have fragile X syndrome—see earlier).

Etiology:

- Other than those with fragile X syndrome, the etiologies are unknown. a. Very strong belief that most autism develops as a result of a multifactorial etiology.
- Genetic basis for some cases is likely, but other than in the case of fragile X, no genetic markers have been identified.
- Maternal ingestion of valproic acid or thalidomide during pregnancy increases the child's risk of developing autism.

Pathophysiology:

- Wide range of pathology, with many variations, including any or all of the following behaviors:

1. Inability to understand and engage in normal social interactions.
2. Inability to form any meaningful relationships, including with the child's own parents.
3. Inability to communicate effectively.
4. Inability to engage in any type of play activities.

- Oft en, the child develops normally, then abnormal behaviors appear when the child reaches 2 to 3 years of age.
- Signs and symptoms vary widely.

1. Social impairment (e.g., ignores the existence of others, plays alone, either doesn't seek comfort when injured or doesn't even acknowledge an injury when it occurs).
2. Language impairment (e.g., fails to engage in conversations or monopolizes conversations with topics of interest only to themselves; exhibits fl at or inappropriate affect when speaking; or fails to engage in any interactive play).
3. Behavioral impairment (e.g., engages in repetitive behaviors, such as hand flapping or head banging; has tantrums over minor changes in the environment or in daily routines; obsesses about following detailed schedules; engages in self-mutilation; exhibits marked sensitivity to sounds or light).
4. Cognitive impairment: most have distinct cognitive defi cits, although the same child may also have areas of marked intelligence (e.g., "Rain Man").

Diagnosis:

1. Screening tool.

a. Th e American Academy of Pediatrics recommends that all children be screened for ASD between the ages of 18 and 30 months using the Modified Checklist for Autism in Toddlers, Revised (M-CHAT-R) (2009).

b. Clinical picture: (the diagnostic signs are usually evident by the age of 3) as defined in the DSM-V.

Treatment:

1. Early diagnosis is essential, with interventions designed for each child's specific needs.
2. Behavioral therapies are most frequently employed.
3. Lifelong intervention is oft en required; autistic individuals have normal life spans.

Nursing Interventions:

1. Deficient Knowledge/Anxiety/Fear/Anger/ Grieving/Impaired Growth and Development.

- Educate the parents and others regarding the child's diagnosis.
- Allow the parents to express grief, anger, and/or frustration regarding the child's diagnosis.
- At each well-child visit, the child's growth and development should be assessed. d. Report any deviations from normal to the primary health-care provider.

2. Impaired Social Interaction/Impaired Verbal Communication (especially important during hospitalizations).

- Have the same nurse care for the child as much as possible or, if the child ever needs a babysitter, encourage the parents to employ the same person each time.
- Encourage the parents to establish a strict routine at home and maintain the routine that is as close to the child's normal as possible during hospitalizations.
- Use alternate means of communication (e.g., pictures) as a way to interact with the child.
- Strongly encourage a family member to accompany the child at all times.
- Refer the child and family to educational resources specifically geared to autistic children.
- Refer the child and family to community resources (e.g., American Autism Society, American Autism Association).

3. Risk for Injury.

- Maintain as safe an environment as possible.
- Provide the child with constant supervision.
- Provide the child with safety equipment when needed (e.g., helmet for head banging).