Unit 2: AUTISM SPECTRUM DISORDER (ASD) & DOWN SYNDROME

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AUTISM SPECTRUM DISORDER (ASD)

DEFINITION

- a condition related to brain development, impacting how individuals perceive and interact with others. Key characteristics include challenges in social interaction, communication, and repetitive behaviors. The "spectrum" reflects the vast range of symptoms and severity. ASD encompasses previously separate diagnoses like autism, Asperger's, and others. Emerging in early childhood, it can eventually hinder social, academic, and professional functioning. Symptoms often appear in the first year, though rarely, regression might occur between 18-24 months.

ETIOLOGY

- Autism spectrum disorder has no single known cause. Given the disorder's complexity and the fact that symptoms and severity vary, there are likely multiple causes. Both genetics and environment may play a part:
    - **Genetics**: Several genes appear to be involved in autism spectrum disorder. A genetic disorder, such as Rett syndrome or Fragile X syndrome can accompany autism spectrum disorder. Other children's genetic changes (mutations) may raise their risk of autism spectrum disorder. Other genes may influence brain development, how brain cells communicate, or the intensity of symptoms. Some genetic mutations appear to be inherited, while others develop spontaneously.
    - **Environmental factors**: Researchers are currently investigating whether factors such as viral infections, medications or complications during pregnancy, or air pollutants play a role in triggering autism spectrum disorder.

PREVALENCE & INCIDENCE

Locally	*Internationally*
● As of March 2023, around 1 in 100 Filipinos, totaling approximately 1.2 million individuals, are living with Autism Spectrum Disorder (ASD) in the Philippines (PNA, 2023). ● Over the last decade, there has been a consistent increase in the prevalence of ASD cases in the country. According to the Department of Health (DOH) statistics, the number of Filipinos diagnosed with ASD has surged from 500,000 in 2008 to 1 million in April 2018. ● The Philippines has autism rates of 81.8 per 10,000 people or 1 in 122 people. However, an updated and precise figure for current ASD cases in the Philippines is yet to be released. ● Data from January 2017 to March 2023 showed that the number of congenital/inborn-Autism totaled 15,901 individuals in which CALABARZON (Region IV) had the most cases.	● As of November 2023, global statistics indicate that approximately 1 in 100 children worldwide is diagnosed with autism. ● The prevalence of Autism Spectrum Disorder (ASD) globally stands at around 75 million individuals, constituting approximately 1% of the world's population. ● Notably, there has been a significant 178% increase in autism prevalence since the year 2000. ● In the United States, it is estimated that 1 in 36 children is affected by autism, with boys being four times more likely than girls to receive a diagnosis (CDC, 2023). ● The prevalence of autism varies widely across countries, from as low as 1 in 250 people in India to as high as 1 in 36 people in the United States (CDC, 2023).

SIGNS, SYMPTOMS, PATHOMECHANICS

Manifestations that the **Physician/Allied Health Professional** Perceive	**Social Communication and Interaction** ● <u>Verbal Communication</u> ○ Delayed speech development ○ Echolalia (repeating others' words) ○ Scripting (repeating memorized phrases)

	○ Difficulties with word order, grammar, or fluency ○ Challenges understanding abstract language or sarcasm ● Nonverbal Communication ○ Infrequent or inconsistent eye contact ○ Unusual body language, posture, or gestures ○ Limited or inappropriate facial expressions ○ Monotone voice or unusual voice prosody **Social Understanding and Interaction** ● Difficulty understanding emotions and intentions ● Challenges regulating own emotions and expressing them appropriately ● Difficulty developing and maintaining friendships ● Lack of interest in or understanding of social norms and cues ● Trouble initiating or maintaining conversations ● Misinterpreting social situations, leading to anxiety or confusion **Repetitive Movements (Stimming)** ● Hand flapping, rocking, spinning, pacing, jumping ● Body rocking, head banging, toe walking ● Stimming behaviors like twirling objects, snapping fingers, or making repetitive sounds **Insistent Routines and Rituals** ● Rigid adherence to routines and schedules ● Extreme distress over minor changes in routine or environment ● Insistence on specific objects or arrangements ● Compulsive behaviors or rituals **Intense or Focused Interests** ● Deep absorption in specific topics ● Obsessive knowledge acquisition and repetitive questioning ● Difficulty shifting attention to other areas of interest

	● Unusual intensity of interest that may seem excessive **Developmental Delays** ● Early language delays (speaking first words after 2 years old) ● Delayed development of social skills (e.g., sharing, turn-taking) ● Motor skill delays (e.g., walking, manipulating objects) **Potential Medical Conditions and Conditions Associated** ● Gastrointestinal (GI) issues: Constipation, diarrhea, food sensitivities ● Sleep disturbances: Difficulty falling asleep, staying asleep, frequent awakenings ● Seizures: Epilepsy or other seizure disorders ● Anxiety and depression: More common in individuals with ASD ● Attention deficit hyperactivity disorder (ADHD): Frequently co-occurs with ASD ● Intellectual disability: Varies greatly across individuals with ASD ● Sensory processing differences: Hypersensitivity or hyposensitivity to sensory input
Manifestations that the **Parents/Significant Others** Perceive	**Social Interaction and Communication** ● Delayed or atypical language development: Parents might notice delays in speech onset, unusual language patterns (echolalia, scripting), difficulty initiating or responding to conversations, or a limited vocabulary compared to peers. ● Social challenges: They might observe struggles with understanding social cues, making eye contact, sharing interests, engaging in pretend play, or initiating playdates. ● Unusual sensory processing: Parents may see their child displaying oversensitivity or undersensitivity to specific sounds, textures, lights, or tastes, leading to distress or avoidance behaviors. ● Restricted interests: Parents may notice intense focus on specific topics or activities, leading to limited exploration of other interests and difficulty transitioning between activities. **Emotional and Behavioral Manifestations** ● Meltdowns and tantrums: Parents often witness meltdowns triggered by sensory overload,

	changes in routine, or communication difficulties. ● Repetitive behaviors: They might observe repetitive movements like flapping, rocking, or spinning, or strong preferences for routine and sameness, which can impact daily activities ● Anxiety and stress: They might observe signs of anxiety related to social situations, unfamiliar environments, or sensory triggers. ● Sleep difficulties: Difficulty falling asleep, staying asleep, or unusual sleep patterns might be evident to parents. ● Feeding challenges: Selective eating, unusual food preferences, or sensory aversions to certain textures or tastes might be observed.
Manifestations that the **Patient** Experiences	**Sensory Processing** ● Oversensitivity: individuals with ASD often describe experiencing certain sounds, lights, textures, smells, or tastes as overwhelming or even painful. This can lead to discomfort, anxiety, and withdrawal from stimulating environments. ● Undersensitivity: Conversely, some may have diminished sensitivity to sensory input, seeking out intense experiences or failing to register certain stimuli. This can manifest as a need for repetitive movements or seeking out stimulating activities. **Social Interaction and Communication** ● Difficulty understanding social cues: individuals with ASD may struggle to interpret facial expressions, body language, and unspoken social rules, leading to confusion and feelings of isolation. ● Challenges with expressing oneself: They may find it difficult to articulate their thoughts and feelings clearly, leading to frustration and misunderstandings. ● Internal world vs. external world: Some individuals with ASD describe feeling like they experience the world differently, leading to a sense of being "out of sync" with others. **Internal Thoughts and Emotions** ● Intense interests and focus: Many individuals with ASD have deep and focused interests

	that bring them joy and engagement. However, the intensity can make it challenging to engage in other activities or social interactions. ● Repetitive thoughts and behaviors: Some may experience repetitive thoughts or engage in repetitive behaviors as a way to cope with anxiety, regulate emotions, or find comfort in predictability. ● Anxiety and stress: The challenges of navigating social situations, sensory overload, and communication difficulties can contribute to anxiety and stress for many individuals with ASD.
Structural & Anatomical Changes	**Brain Volume and Size** ● Early childhood: Studies have shown increased brain volume in young children with ASD compared to typically developing children. This increase often normalizes or slows down later in development. ● Specific regions: Different brain regions show varied volume changes in ASD. Some studies suggest smaller amygdalae (involved in emotion processing) and hippocampi (involved in memory), while others report larger cerebellar volumes (involved in motor coordination and cognition). **Cortical Thickness and Folding** ● Abnormalities in cortical thickness: Regions like the frontal, temporal, and parietal lobes may show atypical thickness patterns in individuals with ASD compared to controls. ● Increased gyrification: Studies suggest some areas of the cortex might have more folds and wrinkles in ASD brains, potentially impacting information processing and connectivity. **White Matter Connectivity** ● Disrupted connections: White matter tracts, responsible for communication between brain regions, may show altered connectivity in ASD individuals. This could affect language processing, social cognition, and other functions.

POSSIBLE SPEECH-LANGUAGE PROBLEMS ASSOCIATED WITH THE CONDITION

All people with autism have some degree of challenge with communication:

Social Aspects of Communication

- Challenges relating to others
- Seeming like they are not interested in others or in making friends
- They often find it hard to have conversations (ASHA, 2023)
- Most have a reduced ability to repair breakdowns in conversation (Rhea Paul, 2018)
- Hard to notice social cues
- Have some type of restricted interests or repetitive behaviors
- It may be hard for a person with autism to
    - Share attention with someone else and focus on the same object or event;
    - Join in play with others and share toys;
    - Respond when others invite them to play or talk;
    - Understand how others feel;
    - Take turns in play or conversation; and
    - Make and keep friends.
- Behaviors of those with ASD:
    - Repeat certain behaviors, including hand or body movements;
    - Cry, laugh, or become angry for unknown reasons;
    - Have trouble changing from one activity to the next;
    - Get upset by certain sounds, smells, or textures;
    - Choose foods based on look or texture; and
    - Show interest in only a few objects or topics.

Receptive and Expressive Language

- Communication skills are limited by odd and tangential speech and repetitive language (Rhea Paul, 2018)

Hearing

- Studies hint that hearing problems are at least three times as common in autistic people as in typical people (Madhusoodanan, 2020) so a hearing test is recommended.

Cognitive Aspects of Communication

- Autism spectrum disorder (ASD) is associated with neurocognitive impairment, including executive dysfunctioning and social cognition (SC) deficits.

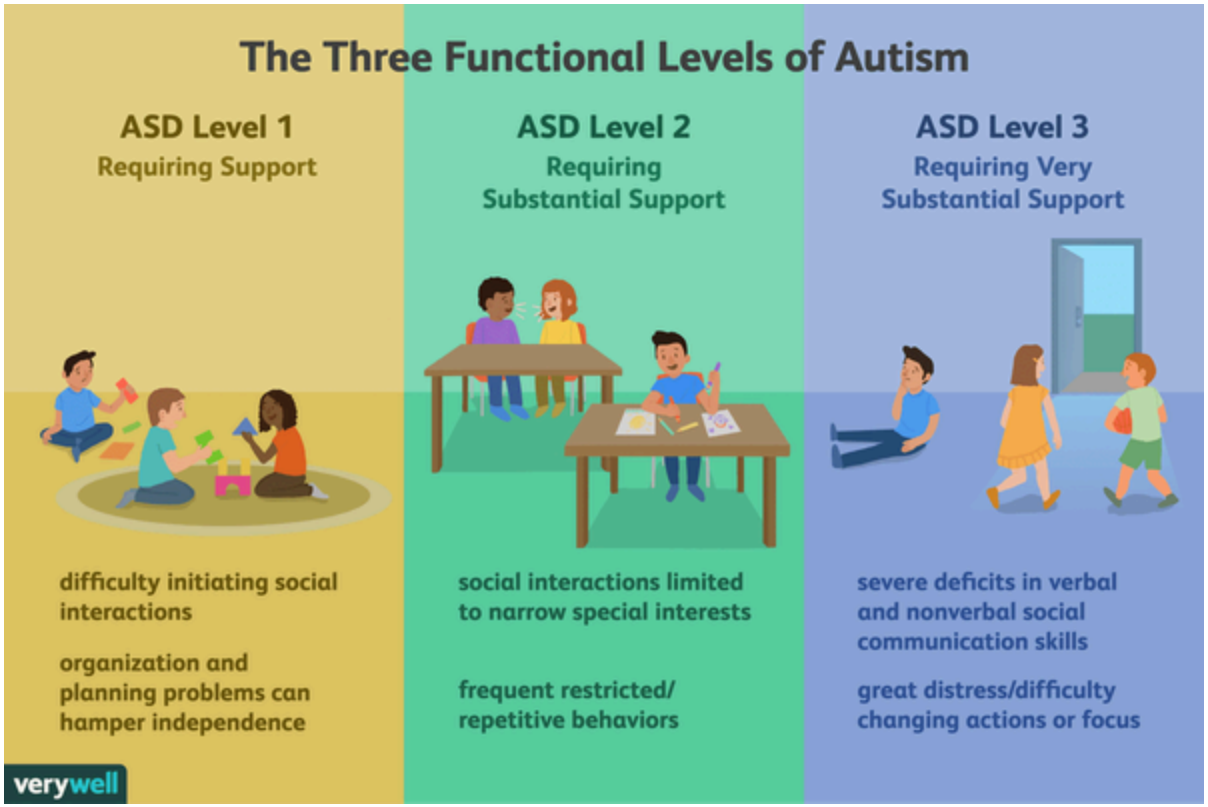
Communication Modalities

- Some people with autism don't talk at all, talk very little, or have trouble talking. SLPs may test the person's ability to use augmentative and alternative communication (AAC) to help them communicate.

TYPES, COURSE, & PROGNOSIS

Types of ASDs	Before 2013, healthcare professionals categorized autism into specific types. 1. **Kanner's Syndrome**, also known as classic autistic disorder, characterizes children as appearing smart and attentive, with traits including an inability to form emotional attachments, uncontrollable speech, obsession with handling objects, communication difficulties, and a high level of rote memory and visuospatial skills, coupled with significant learning problems in other areas. 2. **Asperger's syndrome,** previously identified before 2013, was reclassified as level I ASD by DSM-5. Young individuals with Asperger's syndrome displayed effective communication and academic performance but struggled with social connections. Their behavior and thought patterns tended to be inflexible and repetitive. 3. **Childhood Disintegrative Disorder (CDD),** a rare condition merged into ASD in the Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition, had an onset varying among individuals, typically observed after three years of age. Due to its late onset, children experienced a loss of previously acquired skills in social, verbal, and motor functioning. The progression into CDD often occurs between three and four years but could happen any time before 10 years of age. 4. **Rett Syndrome**, affecting 1 in every 10,000 women and rarely men, was usually diagnosed in children aged 6 to 18 months. It was associated with the failure of developmental milestones or loss of previously acquired abilities. Experts now consider Rett syndrome part of a disease spectrum caused by mutations in the MECP2 gene, involving duplication on the X chromosome and causing serious deficits in various aspects of a child's life. 5. **Pervasive Developmental Disorder-Not Otherwise Specified (PDD-NOS)** was the diagnosis for children with learning and behavioral difficulties not meeting all diagnostic criteria for a specific autism category. Individuals in this group were thought to have milder symptoms than those with ASD. Nevertheless, in 2013, the American Psychiatric Association underwent a revision of its Diagnostic and Statistical Manual of Mental Disorders (DSM-5), resulting in the exclusion of these subtypes of autism. Instead,

	all these distinct categories were unified under the comprehensive umbrella term of Autism Spectrum Disorder (ASD). **Updated Levels:** ● **ASD Level 1** – Level 1 ASD is currently the lowest classification. Those on this level will **require some support** to help with issues like inhibited social interaction and lack of organization and planning skills. ● **ASD Level 2** – In the mid-range of ASD is Level 2. At this level, individuals **require substantial support** and have problems that are more readily obvious to others. These issues may be trouble with verbal communication, having very restricted interests, and exhibiting frequent, repetitive behaviors. ● **ASD Level 3** – On the most severe end of the spectrum is Level 3 which **requires very substantial support**. Signs associated with both Level 1 and Level 2 are still present but are far more severe and accompanied by other complications as well. Individuals at this level will have limited ability to communicate and interact socially with others.

	
Course	The prognosis of a child with autism depends on how severe their symptoms are initially, but early intervention and treatment can make a positive difference. While autism was once believed to be irreversible, there are now effective treatments that support individuals throughout their lives. Autism may come with accompanying conditions, and addressing these can improve the outlook of their progress. Autism usually starts in early childhood and can create challenges in social, school, and work situations. As children with autism grow up, some become more engaged and show fewer behavior issues, while others may continue to struggle with language or social skills, especially during the teenage years.

	Children with autism typically face difficulties in social skills, communication, motor skills, and sensory processing. These challenges can make it hard for them to interact with peers and follow instructions in school. With this, early intervention is crucial for better outcomes. In recent years, there have been more children diagnosed with autism attending regular school and living somewhat independently. However, most individuals with autism still experience some difficulties in communication and socialization. In the field of mental health, adults with autism experience higher rates of co-occurring conditions compared to the general population. Around 54% of adults with Autism Spectrum Disorder (ASD) have at least one psychiatric condition, such as anxiety, mood disorders, psychotic disorders, and more. Behavioral challenges, such as being disruptive, engaging in self-harm, or destructive behavior, are prevalent among 46% of autistic adults. Furthermore, the occurrence of mental health issues in adults with autism tends to rise with age.
Outcome if Left Treated and/or Untreated	Detecting autism early in life creates an opportunity for early interventions, which can provide essential support while nurturing the distinctive skills of the individual. When Autism Spectrum Disorder (ASD) is left untreated, misdiagnosed, or diagnosed late, the negative symptoms linked to the condition may deteriorate over time. Without adequate support, children may struggle to develop crucial skills in learning, speech, and social interactions. Adults who haven't received proper treatment may encounter difficulties in living independently, finding employment, and maintaining relationships. The comprehensive goals of autism treatment include addressing developmental delays, improving social understanding, enhancing communication and learning capacities, promoting mood regulation, teaching self-soothing techniques, and fostering existing skills.

HEALTHCARE RESOURCES AVAILABLE FOR ASD

Screening & Diagnosis of ASD	
Developmental **Monitoring**	● An active, ongoing process of watching a child grow and encouraging conversations between parents and providers about a child's skills and abilities. ● Involves observing how the child grows and whether they meet the typical developmental milestones, or skills that most children reach by a certain age, in playing, learning, speaking, behaving, and moving.

Developmental **Screening**	● More formal than developmental monitoring ● Involves screening questionnaires and checklists based on research that compares them to their same-age peers
Developmental **Diagnosis**	● A formal developmental evaluation ● A more in-depth look at a child's development is usually done by a trained specialist such as a developmental pediatrician, child psychologist, speech-language pathologist, occupational therapist, or other specialist.

Treatment & Intervention Services	
Behavioral Approaches	● Focus on changing behaviors by understanding what happens before and after the behaviors
Developmental Approaches	● Focus on improving specific developmental skills, such as language skills or physical skills, or a broader range of interconnected developmental abilities
Educational Approaches	● Given in a classroom setting ● E.g. Treatment and Education of Autistic and Related Communication-Handicapped Children
Social-Relational Approaches	● Focus on improving social skills and building emotional bonds; some involve parents or peer mentors
Pharmacological Approaches	● There are no medications that treat the core symptoms of ASD but some treat co-occurring symptoms that can help people with ASD function better
Psychological	● Help people with ASD cope with anxiety, depression, and other mental health issues

Approaches	
Complementary & **Alternative** Treatments	● Treatments that do not fit into any of the other categories ● Used to supplement more traditional approaches

Accessing Services	
Early Intervention Services (Ages 0-3)	● Can greatly improve a child's development and result in better outcomes ● Help children from birth to 3 years old learn important skills
Special Education Services (Ages 3-22)	● Children with ASD may be eligible for services beginning at age 3 years ● They often have an individualized education plan

SLP THERAPY & EVALUATION

SLP Areas	*Materials*	*Strategies*
Social/Pragmatic Communication Development	**Pragmatic Language Assessment Tools** ● Pragmatic Language Skills Inventory (PLSI) ● Test of Pragmatic Language (TOPL) ● Clinical Evaluation of Language Fundamentals ● Social Language Development Test	**Strategies & Approaches** ● Roleplay ● Turn-taking games ● Play dates ● Facial expressions ● Miming ● Describing activities ● Puppets ● Comic strips ● Social skills groups ● Social stories ● Greetings
Receptive Language	**Receptive Language Assessments** ● Assessment of Comprehension and Expression (ACE) ● New Reynell Developmental Language Scales (NRDLS) ● York Assessment of Reading for Comprehension (YARC) ● Wechslet Individual Achievement Test (WIAT-III) ● Clinical Evaluation of Language Fundamentals (CELF) ● Test for Reception of Grammer (TROG) ● Test of Abstract Language Comprehension	**Strategies & Approaches** ● Eye-contact ● Minimal Instructions ● Simplifying Language ● Chunking Verbal Language into Parts ● Repeat ● First/Then ● Clarify ● Show ● Visual Aids ● Describe ● Emphasizing Words ● Play

	(TALC)	• Reducing Background Noise • Face-to-Face • Visual • Books
Expressive Language	**Expressive Language Assessments** • Assessment of Comprehension and Expression (ACE) • Wechsler Individual Achievement Test (WIAT-III) • Clinical Evaluation of Language Fundamentals (CELF) • Preschool Language Scales (PLS) • New Reynell Developmental Language Scales (NRDLS) • Bayley Scales	**Strategies & Approaches** • Play • Talking to the child • Turning off background noise • Face-to-face • Expanding the language • Books • Modeling Back • Choice-Making • Singing Songs • Using pictures/drawings/photos
Theory of Mind	**Theory of Mind Assessments** • False Belief Tasks • Sally-Anne Test • Theory of Mind Task Battery	**Strategies & Approaches** • Using Mental State Verbs in Daily Routine • Linking Concrete Objects with Mental State Verbs • Talking About Past Experiences • Talking About Upcoming Events • Naming Mental States as They Happen

THE HEALTHCARE TEAM FOR ASD

BEHAVIORAL TREATMENT	*DEVELOPMENTAL TREATMENT*	*EDUCATIONAL TREATMENT*	*PHARMACOLOGICAL TREATMENT*	*ALTERNATIVE TREATMENT*
Psychologists	Speech-Language Pathologists	Special Educators	Physicians	Dieticians
Psychiatrists	Occupational Therapists	Resource Teachers	Pediatricians	Art Therapists
Counsellors	Physical Therapists			Others

SUPPORT SYSTEMS FOR PEOPLE WITH ASD

Autism Partnership Philippines (APP)

- Autism Partnership Philippines officially commenced its operations in March 2015 in Quezon City, Manila, making it the first office in the Philippines and part of a network that includes locations in Asia such as Hong Kong, Singapore, and Korea. The center, known as AP Philippines, has become one of the well-established and promising facilities providing professional Applied Behavior Analysis (ABA) therapy services to families for the treatment of individuals with Autism Spectrum Disorder.
- APP boasts a Clinical Team in the Philippines consisting of behavioral therapists, program supervisors, and consultants. These consultants extend their expertise globally by providing overseas training and workshops to families, schools, organizations, and related agencies. Their outreach spans countries like Malaysia, Indonesia, Vietnam, China, and Kuwait.

The Autism Society Philippines (ASP)

- The Autism Society Philippines (ASP) is a national, non-profit organization with a mission to create an environment that empowers individuals on the autism spectrum to reach their full potential—becoming self-reliant, independent, productive, and socially-accepted members of an Autism-OK Philippines. ASP is dedicated to establishing institutional mechanisms that support individuals on the autism spectrum and their families.
- ASP focuses on designing programs to support families and individuals on the autism spectrum. ASP facilitates regular family support group meetings, sibling support, workshops, seminars, and training covering various topics. Their services include early detection and intervention consultancy, free community-based therapy, and the sharing of scientific updates. ASP,

actively engaged in autism advocacy, organizes monthly seminars globally, emphasizing education and therapy partnerships for the development of individuals on the autism spectrum. Additionally, ASP takes a lead role in various celebrations during the National Autism Consciousness Week and World Autism Awareness Day in the Philippines.

DOWN SYNDROME

DEFINITION

- Down syndrome (trisomy 21), is a genetic disorder caused when abnormal cell division results in an extra full or partial copy of chromosome 21. It is the most common chromosomal disorder and leading cause of learning disabilities, characterized by varying degrees of intellectual disability, distinctive physical features, and an increased risk of medical conditions like heart defects.

ETIOLOGY

- Down syndrome is caused by a random error in cell division, which results in an extra copy of chromosome 21. This type of error is known as nondisjunction. Typically, when one cell divides into two, pairs of chromosomes are separated so that one of the pairs travels to one cell and the other to the other. Nondisjunction occurs when something goes wrong and both chromosomes from one pair enter one cell but not the other.

PREVALENCE & INCIDENCE

Locally	*Internationally*
● One in every 800 babies born in the Philippines has Down syndrome (PNA, 2023). ● According to DSAPI or the Down Syndrome Association of the Philippines Inc., there are approximately 1,875 babies born with Down syndrome each year. ● This brings about 100,000 households living with a person with Down syndrome. ● A Filipino with Down syndrome is born in the country every four hours (Punay, 2015).	● The estimated incidence of Down syndrome is between 1 in 1,000 to 1 in 1,100 live births worldwide. Each year, approximately 3,000 to 5,000 children are born with this chromosome disorder (UN, 2023). ● In 2015, Down syndrome was present in 5.4 million individuals globally and resulted in 27,000 deaths, down from 43,000 deaths in 1990 (Lancet, 2015). ● In the United States, about 6,000 babies are born with Down syndrome each year, which is about 1 in every 700 babies born. The prevalence of Down syndrome increases as the mother's age increases. Between 1979 and 2003, the number of babies born with Down

	syndrome increased by about 30%. In 2008 about 250,700 children, teens, and adults were living with Down syndrome in the United States (CDC, 2023). ● Ireland has the highest number of babies with Down syndrome – 27.5 out of 10,000 born there (Whitten, 2023).

SIGNS, SYMPTOMS, AND PATHOMECHANICS

Manifestations that the **Physician/Allied Health Professional** Perceive	**Physical Characteristics** ● Flattened facial profile with a small nose and a protruding tongue ● Upward slanted eyes with epicanthic folds (skin folds at the inner corner of the eyes) ● Small ears set low on the head ● Short neck ● Short stature with small hands and feet ● Single palmar crease on the hand ● Wide gap between the big and second toes **Social Communication and Interaction** ● <u>Verbal communication</u> ○ Delayed speech development and articulation difficulties. ○ Limited vocabulary and grammatical complexity. ○ Difficulty understanding abstract concepts and figurative language. ○ May struggle with turn-taking and staying on topic in conversations. ● <u>Nonverbal communication</u> ○ Difficulty interpreting nonverbal cues of others, leading to misunderstandings. ○ Limited use of gestures to elaborate on verbal communication. **Social Understanding and Interaction**

	• Difficulty understanding social cues: This can lead to misinterpretations and social awkwardness. • Difficulty understanding sarcasm, humor, and nonverbal communication. • Possible shyness or hesitancy in social situations • Strong desire for connection and friendship: Individuals with Down syndrome form deep bonds and thrive on positive social interactions. **Repetitive Movements (Stimming)** • Common manifestation: Stimming behaviors like hand flapping, rocking, or body tapping serve various purposes for self-regulation, communication, and managing sensory **Insistent Routines and Rituals** • Anxiety and distress with disrupted routines. • Behavioral outbursts due to unexpected changes. **Intense or Focused Interests** • Difficulty engaging in other activities and socializing. • Interests becoming all-consuming. **Developmental Delays** • Delayed motor skills, such as sitting, crawling, and walking • Delayed speech and language development • Learning difficulties and cognitive delays • Difficulty with social interaction and communication **Potential Medical Conditions and Conditions Associated** • Congenital heart defects: The most common medical issue, occurring in about half of babies born with Down syndrome. Early diagnosis and treatment are critical. • Hearing loss: Can affect speech and language development. Regular hearing checks and appropriate interventions are necessary.

	● Vision problems: Common issues include nearsightedness, farsightedness, and cataracts. Eye exams and corrective lenses are important. ● Thyroid problems: Hypothyroidism (underactive thyroid) is more common in individuals with Down syndrome. Regular monitoring and medication may be needed. ● Gastrointestinal issues: Constipation, celiac disease, and gastroesophageal reflux disease (GERD) are more likely. Dietary adjustments and medical management can help. ● Sleep apnea: Interrupted breathing during sleep can lead to fatigue and other health problems. Treatment options include lifestyle changes and breathing assistance devices. ● Increased risk of leukemia: Early detection and treatment are crucial. ● Alzheimer's disease: Dementia is more likely in later life, but there are measures to promote cognitive well-being.
Manifestations that the **Parents/Significant Others** Perceive	**Social Interaction and Communication** ● Limited or delayed language development: Reduced babbling, echolalia, limited vocabulary, difficulty understanding simple requests. ● Reduced or atypical social interaction: Less eye contact, delayed social smile, preference for solitary play, difficulty initiating interaction. ● Non-verbal communication challenges: Limited gestures or facial expressions, difficulty understanding and using gestures appropriately. ● Difficulty understanding emotions: Trouble recognizing and responding to emotions in others. ● Difficulty making and keeping friends: Challenges understanding social cues and body language, trouble initiating and maintaining conversations. ● Social inflexibility: Difficulty adapting to changes in routine or social situations, may display shyness, anxiety, or frustration. ● Expressive language delays: Limited vocabulary compared to peers, difficulty with grammar and sentence structure, trouble following complex instructions. ● Speech difficulties: Articulation challenges, unclear or monotone speech, limited vocal

	inflection. ● Literal interpretation: Difficulty understanding jokes, sarcasm, or figurative language. **Emotional and Behavioral Manifestations** ● Increased sensitivity or insensitivity: Some children might be hypersensitive to noise, touch, or certain textures, while others might seem under-responsive to stimuli. ● Repetitive behaviors: Parents might observe self-stimulatory behaviors like hand-flapping, rocking, or lining up objects, which can serve as self-soothing mechanisms. ● Strong attachments and affection: Children with Down syndrome often form strong bonds with their caregivers and show their love and affection in unique ways.
Manifestations that the **Patient** Experiences	**Sensory Processing** ● Sensitive to textures, sounds, and environments: Some individuals may experience sensory sensitivities similar to those with ASD, finding certain stimuli overwhelming or unpleasant. This can lead to withdrawal, discomfort, or seeking out calming environments. ● Undemanding sensory preferences: Others may not have significant sensory sensitivities, simply favoring familiar surroundings and routines for comfort and predictability. **Social Interaction and Communication** ● Challenges understanding social cues: Similar to individuals with ASD, understanding nonverbal communication like facial expressions and body language can be difficult, leading to social confusion or awkwardness. ● Social desire and motivation: Despite communication challenges, many individuals with Down syndrome have a strong desire for social interaction and connection. They may express this through affection, shared activities, or simply being present with others. ● Difficulties with expressive language: Limited vocabulary and articulation skills can create barriers in communication, requiring patience, alternative methods (gestures, pictures), and

	understanding from others. **Internal Thoughts and Emotions** ● Varying degrees of intellectual understanding: Individuals with Down syndrome experience the world with different levels of understanding, depending on the severity of their cognitive limitations. This can lead to frustration or confusion if their internal world doesn't match external expectations. ● Limited vocabulary and expression: Language barriers can limit their ability to articulate thoughts and feelings, potentially causing emotional isolation or miscommunication. ● Positive personality traits: Many individuals with Down syndrome are described as friendly, affectionate, and cooperative, experiencing joy and love in their own way. ● Potential for anxiety and depression: Social difficulties, health concerns, and communication barriers can contribute to anxiety and depression, requiring emotional support and coping strategies.
Structural & Anatomical Changes	**Craniofacial** ● Flattened facial profile: This includes a smaller-than-average nose with a flat bridge, upslanting eyes with epicanthic folds (skin folds), and a small lower jaw. ● Short neck: This can contribute to sleep apnea and breathing difficulties. ● Small ears: Often low-set and with a curved-over top. **Skeletal Features** ● Single palmar crease: This crease runs across the center of the palm instead of having the typical three. ● Short stature: Individuals with Down syndrome tend to be shorter than average. ● Short, broad hands and feet: Hands may have short fingers and a gap between the first and second toes. ● Hypotonia: Decreased muscle tone can affect motor skills and coordination. ● Joint hypermobility: Joints may be looser than usual, increasing the risk of dislocations.

	Internal organs ● Congenital heart defects: About half of individuals have heart defects present at birth. ● Gastrointestinal issues: These can include blockages in the digestive system, celiac disease, and gastroesophageal reflux disease (GERD). ● Hearing and vision problems: Hearing loss and nearsightedness are common. ● Thyroid issues: An underactive thyroid (hypothyroidism) is seen in around 15% of cases. **Brain** ● Smaller brain size: This can lead to cognitive and learning challenges. ● Simplified gyral pattern: Fewer folds and grooves on the brain's surface. ● Hippocampal abnormalities: The hippocampus plays a role in memory and learning, and its structure and function may be affected.

POSSIBLE SPEECH-LANGUAGE PROBLEMS ASSOCIATED WITH DOWN SYNDROME

Children and adults with Down Syndrome are impacted by difficulties with hearing, feeding, speech and language.

Articulation

- Speech intelligibility is one of the most difficult areas for people with Down syndrome at all ages.
- Articulation problems occur with specific sounds. Children with Down syndrome are slow to acquire the phonological system of their mother tongue (Stoel, 2001).

Voice and resonance

- Low vocal pitch and hoarse, harsh or raucous voice have frequently been ascribed to individuals with Down syndrome (Kent & Vorperian, 2012).

Receptive and expressive language

- People with Down syndrome frequently have difficulty with grammar, tenses and word endings and use shorter sentences to communicate.

- They have a receptive-expressive gap—their test scores for receptive language are higher than for expressive language.

Hearing

- Anatomic differences in the ears of children with Down syndrome (narrow and short canals) make it more susceptible to accumulations of fluid behind the eardrum causing a hearing loss condition called Otitis Media with Effusion (OME) (Matthews, 2013).
- Children with Down syndrome often have fluctuating hearing loss due to the frequency of fluid accumulation.

Swallowing

- Low muscle tone (hypotonia) affects feeding.
- Difficulty with sensory processing and oral tactile feedback affect swallowing.
- People with Down syndrome may experience issues like silent aspiration, reduced mouth alertness for feeding, swallowing air leading to stomach discomfort, loss of interest in eating, and difficulties transitioning to spoon feeds and tolerating various textures.

Cognitive aspects of communication

- People with ASD have difficulties in motor planning for speech
- They often have mild to moderate intellectual disability and may have specific challenges with attention span, verbal memory, and expressive communication.
- Behavioral problems such as stubbornness, impulsivity, and temper tantrums may be more common.

Social aspects of communication

- Children with Down syndrome are at a disadvantage in establishing relationships because of their cognitive and communicative difficulties, often missing out on early opportunities to engage in social interactions with peers (Poikey, 2018).
- Have a basic understanding of language for social interaction, but they often show significant pragmatic weaknesses, particularly in abstract contexts (Abbeduto, 2008).

Communication modalities

- Alternative Augmentative Communication (AAC) can offer great benefits for individuals with Down Syndrome when it is needed. It aids language development, fostering language use, boosting communication confidence, and facilitating the development of social language and relationships.

TYPES, COURSE, PROGNOSIS

Types of Down Syndrome	There are different kinds of Down syndrome, and they all involve having an extra piece of the 21st chromosome. Here are the main types:: 1. **Trisomy 21 Down Syndrome:** This is the most common type, making up about 95% of cases. It happens when there's a mistake in how the cells divide during the baby's development. Instead of getting one chromosome from each parent, the baby ends up with an extra one in the 21st pair. As the baby grows, this extra chromosome is copied into all its cells. 2. **Translocation Down Syndrome:** In this type, there's still an extra piece of chromosome 21, just like in trisomy 21. But in these cases, the extra piece is attached to one of the other 23 chromosomes. People with translocation Down syndrome have the usual 46 chromosomes, but one of them is connected to an extra piece of chromosome 21. This type is less common, making up only 3 to 4% of cases. 3. **Mosaic Down Syndrome:** Mosaic Down syndrome is the rarest type, accounting for 1 to 2% of cases. In this case, some cells have an extra copy of chromosome 21, but not all of them. People with mosaic Down syndrome might have milder symptoms or fewer issues than those with other types of Down syndrome.
Course	Individuals with Down syndrome may experience various complications and some of these challenges become more noticeable as they age. These complications can include: ● **Heart Defects:** Around half of children with Down syndrome are born with congenital heart defects. These heart issues may be serious and require surgery during early infancy. ● **Gastrointestinal (GI) Defects:** Some children with Down syndrome may have GI abnormalities, affecting the intestines, esophagus, trachea, and anus. This can increase the risk of digestive problems like GI blockage, heartburn, or celiac disease.

	• **Immune Disorders:** Due to abnormalities in their immune systems, individuals with Down syndrome face an elevated risk of developing autoimmune disorders, certain cancers, and infectious diseases such as pneumonia. • **Sleep Apnea:** Children and adults with Down syndrome are at a higher risk of obstructive sleep apnea. Changes in soft tissue and skeletal structures can lead to airway obstruction during sleep. • **Obesity:** People with Down syndrome are more prone to obesity compared to the general population. • **Spinal Problems:** Some individuals with Down syndrome may have a misalignment of the top two neck vertebrae, known as atlantoaxial instability. This condition increases the risk of spinal cord injury from overextension of the neck. • **Leukemia:** Young children with Down syndrome face an increased risk of leukemia. • **Dementia:** There is a significantly heightened risk of dementia for individuals with Down syndrome, with signs and symptoms potentially emerging around age 50. Additionally, the risk of developing Alzheimer's disease is increased. • **Other Problems:** Down syndrome may also be associated with various other health conditions, including endocrine issues, dental problems, seizures, ear infections, and challenges with hearing and vision.
Outcome if Left Treated and/or Untreated	For individuals with Down syndrome, maintaining good health involves regular medical check-ups and addressing issues as they arise. They are more prone to certain health concerns compared to the general population, and many may have lower immunity. Early intervention is crucial, and routine check-ups and screenings play a vital role in promoting a healthy lifestyle. Given the increased susceptibility, it's essential to provide extra care, especially in the early and later stages of life. Prompt attention to common medical problems like colds and infections is important to prevent them from escalating into more serious issues. Without timely treatment, these common health issues could potentially develop into more severe conditions, impacting their daily lives and posing risks to their well-being. Regular monitoring and timely medical care contribute significantly to the overall health and quality of life of

	individuals with Down syndrome.

HEALTHCARE RESOURCES AVAILABLE FOR DOWN SYNDROME

Prenatal Testing for Down Syndrome	
Age-Related Risks	● Understanding how the chance of having a baby with Down Syndrome is related to the pregnant person's age
Screening Tests	● Sequential Integrated Screening
Diagnostic Tests	● Amniocentesis ● Chorionic Villus Sampling (CVS) ● Ultrasound

Treatment & Intervention Services	
Early Intervention	● A range of specialized programs and resources that professionals provide to very young children with Down Syndrome and their families
Educational Therapy	● Appropriate educational services and devices to help children with down syndrome learn as much as they can
Treatment Therapies	● Physical Therapy ● Speech-Language Therapy ● Occupational Therapy ● Emotional & Behavioral Therapies

Drugs & Supplements	● Amino acid supplements or drugs that affect their brain activity ● Psychoactive drugs
Assistive Devices	● Assistive devices—any type of material, equipment, tool, or technology that enhances learning or makes tasks easier to complete

SLP THERAPY & EVALUATION

SLP Areas	*Strategies*
Working Memory	**Intervention Strategies** ● Improve phonological loop function ○ Reducing hearing difficulties ○ Auditory discrimination of speech sounds and words ● Improve remembering lists of items ● Improve attention and increase processing capacity ● Grouping or Organization Skills
Motor Development	**Intervention Strategies** ● Playing with Musical Instruments ● Sorting Activities ● Playing with Dough ● Stickers ● Posting Games ● Building Blocks ● Drawing ● Puzzles ● Threading and Lacing Games ● Cutting

Number Skills	**Intervention Strategies** ● Integration of Computer Software ● Tactile and Visual Strategies ● Using Manipulatives ● Pairing auditory information with visual information ● Providing a variety and abundance of rehearsal and review
RL & EL	*Same as ASD*

THE HEALTHCARE TEAM FOR DOWN SYNDROME

BEHAVIORAL TREATMENT	DEVELOPMENTAL TREATMENT	EDUCATIONAL TREATMENT	PHARMACOLOGICAL TREATMENT
Psychologists	Speech-Language Pathologists	Special Educators	Physicians
Psychiatrists	Optometrist/Hearing Specialist	Resource Teachers	Pediatricians
Counsellors	Occupational Therapists		
	Physical Therapists		

SUPPORT SYSTEMS FOR PEOPLE WITH DOWN SYNDROME

Down Syndrome Association of the Philippines

- The Down Syndrome Association of the Philippines is an organization dedicated to working with children with Down syndrome and their families. With offices nationwide, including main offices in Makati and branch offices in Davao City and Cebu City, they aim to provide support and assistance.

- The Down Syndrome Association of the Philippines offers valuable services in advocacy, raising awareness, family support, and specialized programs for children. They maintain a Facebook Group Page where member families, who have loved ones with Down syndrome, share experiences, seek advice, and build a supportive community.

Disorder Support International (DSI)

- Disorder Support International (DSI) is an organization committed to creating a more inclusive world for those in need. Their mission revolves around fostering a supportive environment for individuals facing various challenges.
- DSI is dedicated to collaborative efforts, providing support to those reaching out for help, and empowering individuals with disabilities to actively participate in global development. Their core beliefs include championing equal human rights, striving for a high quality of life for everyone, breaking the cycle of disability and poverty, and recognizing the benefits of diversity and cross-cultural collaboration.

The Global Down Syndrome Foundation

- The Global Down Syndrome Foundation, established in 2009, is a nonprofit organization dedicated to enhancing the lives of individuals with Down syndrome. The Foundation primarily supports the Linda Crnic Institute for Down Syndrome, the first U.S. academic institution dedicated exclusively to researching and providing medical care for individuals with Down syndrome.
- The Global Down Syndrome Foundation focuses on significantly enhancing the lives of individuals with Down syndrome through research, medical care, education, and advocacy. They actively engage with governments, educational organizations, and society to bring about legislative and social changes, ensuring equitable opportunities for every person with Down syndrome.

Philippine Information Agency

- The Philippine Information Agency (PIA) serves as the official public information arm of the Government of the Republic of the Philippines. Collaborating with the Office of the President, national government agencies, and other public sector entities, PIA communicates its programs, projects, and services to the Filipino people.
- PIA celebrates National Down Syndrome Consciousness Month every February, aiming to educate people about the lives of individuals with Down syndrome. The focus is on dispelling misconceptions and highlighting the unique abilities and fulfilling lives of people with Down syndrome. This includes their participation in schools, showcasing talents, expressing emotions like anyone else, and engaging in various regular activities. PIA emphasizes the importance of celebrating diversity and individuality, in collaboration with the Down Syndrome Association of the Philippines, Inc.

Precautionary and Protective Measures for Clinician Safety

In Speech-Language Pathology, it is crucial to address safe and unsafe scenarios in speech therapy. Creating a mutually supportive and inclusive environment for both the clinician and the client. For sessions with people with ASD and Down Syndrome, these are the different precautionary and protective measures for clinician safety.

Pre-Planning	● Packing/Preparing the right tools ● Setting expectations for arrival ● Assessing environment ● Planning emergency (for home-based services)
General Safety	● Attire Comfortability & Practicality ● Protective Tools ● Safeguarding Privacy
Safety from Physical Injuries, Infections, & Bodily Fluids	● Wearing appropriate PPE ● Environmental Considerations and Preparation ● Animal Considerations
Behavior Management Strategies	● Building Rapport ● Involving Family in Virtual Sessions ● Use of Praise ● Use of Manipulatives ● Use of Aids ● Session Modification ● Environmental Modifications ● Verbal Redirections/Verbal Prompting by Therapist

	• Use of Predetermined consequences • Physical Proximity • Space/Time Out • Physical Activity
Safety from Stress & Burnout	• Mental Health • Considering Professional Help • Advocating for Yourself

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