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Pervasive Developmental Disorders

Introduction

Pervasive Developmental Disorders (PDD), is a term used to refer to a group of neurobiological disorders characterized by fundamental impairments or delays in the development of social interaction skills ,verbal and nonverbal communication skills and motor skills or by the presence of stereotyped (repetitive rudimentary) behaviors . [1]

Based on the International Statistical Classification of Diseases and Related Health Problems 10th **(ICD-10) ,** pervasive developmental disorders are classified into five different conditions :

- Pervasive developmental disorder not otherwise specified (PDD-NOS).
- Childhood Autism, the best-known, now understood to be part of a spectrum;
- Asperger syndrome;
- Rett syndrome; and
- Childhood disintegrative disorder (CDD)[(2)]

In May 2013, the Diagnostic and Statistical Manual-Fifth Edition (DSM-5) was released, updating the classification for pervasive developmental disorders. The grouping of disorders, including PDD-NOS, Autism and Asperger's Syndrome , has been removed and replaced with the general term of Autism Spectrum Disorders. [(3)]

The main focus of this paper is on Autism spectrum disorders (ASD) , Rett syndrome and Childhood disintegrative disorder (CDD) , their characteristics , diagnosis and therapy .

Autism Spectrum Disorders

Definition

Autism spectrum disorders (ASD) is an umbrella term that refers to a group of neurodevelopmental disorders with symptoms appearing in early childhood. As the name indicates, the symptoms are seen on a continuum ranging from mild to severe expression .The three areas of impairment in ASD are reduced to two areas in the DSM-V namely, a social-communication domain and a behavioral domain including fixated interests and repetitive behaviors and a description of the severity level of the ASD .[4]

Autism Spectrum Disorders in the DSM-V

The validity of the DSM-IV was assumed be insufficient, because it included large numbers of diagnoses. Moreover, earlier studies have that there is an overlap between the criteria for Asperger's disorder and autistic disorder and there is an inconsistent distinction between the different ASDs. Thus , the DSM-V characterizes the heterogeneity of the autism spectrum disorders rather than continuing to divide them into sub groups .[4]

Note: Individuals with a well-established DSM-IV diagnosis of autistic disorder, Asperger's disorder, or pervasive developmental disorder not otherwise specified should be given the diagnosis of autism spectrum disorder.

Autism spectrum disorders Diagnostic Criteria in the DSM-V

A. Persistent deficits in **social communication and social interaction** across multiple contexts.

B. **Restricted, repetitive patterns of behavior**, interests, or activities.

C. Symptoms must be present in the **early developmental period** (but may not become fully manifest until social demands exceed limited capacities, or may be masked by learned strategies in later life).

D. Symptoms cause clinically significant **impairment in social, occupational,** or other important areas of current functioning.

E. These disturbances **are not better explained** by intellectual disability (intellectual developmental disorder) or global developmental delay. Intellectual disability and autism spectrum disorder

frequently co-occur; to make comorbid diagnoses of autism spectrum disorder and intellectual disability, social communication should be below that expected for general developmental level.

Causes of Autism spectrum disorders

It is accepted that an interaction of **genetic predispositions and environmental risk factors** play a role in the development of ASD .a brief description of these etiologies is provided below :

1- Gene predisposition : recent analysis of 192 twins with ASD in California reported concordance of 77% for identical male twins and 31 for fraternal male twins .Further research examines family studies , candidate gene studies and association with genetic disorders to provide a linkage between ASD and genetic mutations .

2-Brain anatomy and physiology in ASD : It is reported that 60% of boys with ASD have macrocephaly, with accelerated growth observed between 4 and 12 months of age .Studies using neuroimaging procedures showed functional differences in white matter volume , mirror neurons and theory of mind in the brains of autistic individuals .

3-Obstetric complications : premature infants have a high risk for developing autism .

4-Environmental exposure : teratogens are substances that increase the risk of birth defects in the developing fetus , measles , mumps and rubella (MMR)vaccine , and infections are suggested to be related to ASD .However , no study provides scientific evidence to prove it .

5- Gender and ASD : Autism affects more boys than girls with general ration from 2:1 to 5:1

Diagnostic categories within the autism spectrum disorders

Autistic disorder / **Autism** : This condition was first identified by Dr Leo Kanner (1943) , who studied 11 cases of autistic children. Based on his observations, he concluded that they had profound social aloofness, qualitative differences in language development , language use , remarkably good memory and a desire for preservation of sameness . [(4)(6)]

Kanner implied an organic basis for the disorder [(4)], but also noted that most of autistic children were born to highly intellectual parents . He made an incorrect assumption that the disorder might at least partially result from the cold, intellectual nature of their parents, especially their mothers .In fact , Karl

is attributed with coining the term “refrigerator mother “ which blames mothers for their children's atypical behavior. The theory of “ refrigerator mother” was later disapproved by several research which demonstrated that parents of children with ASD are no difference from parents of other children with disabilities .[7]

Asperger’s disorder: Asperger's syndrome was named for the Austrian doctor, Hans Asperger, who first described the disorder in 1944. He described children with Asperger’s disorder as having apparently typical language, but had repetitive behaviors and difficulties with socialization. He referred to the syndrome as autistic psychopathy.[8]

Children with Asperger's syndrome typically function better than do those with autism. In addition, children with Asperger's syndrome generally have normal intelligence and near-normal language development, although they may develop problems in communications as they get older[9]. There’s an overlap between high functioning autism (autistic disorder in someone with typical intelligence) and Asperger’s disorder.[4]

Diagnostic criteria for Asperger’s disorder are as follows [10]:

- Gross and sustained impairment in social interaction .
- Restricted repetitive & stereotyped patterns of behavior, interests and activities.
- Lack of any significant general delay in language (e.g. single words are used by age 2 years, communicative phrases are used by age 3 years).
- Lack of any clinically significant delay in cognitive development or in the development of age-appropriate self-help skills, adaptive behavior (other than in social interaction) and curiosity about the environment.
- Not better accounted for by another specific pervasive developmental disorder .

Pervasive Developmental Disorder, Not Otherwise Specified (PDD-NOS) is a term used to describe children who do not have the prerequisite number of symptoms or criteria are not met for a specific pervasive developmental disorder, schizophrenia, schizotypal personality disorder, or avoidant personality disorder Significant functional impairment occurs due to the cognitive, intellectual ,language and behavioral deficits associated with PPD-NOS. [10]

Diagnostic features of ASD

1- Qualitative impairments in social reciprocity :

Social reciprocity is the back-and-forth flow of social interaction. The term refers to how the behavior of one person influences and is influenced by the behavior of another person and vice versa. The skills involved in social reciprocity in very young children begin with showing interest in interacting with others and exchanging smiles . Infants learn that eye contact with an adult leads to attention ,. Later , the ability to share a common point of reference is called joint attention starts to develop around 6 months of age . Another skill of social reciprocity is understanding that others have a different point of view(theory of mind) .[4]

Impairments in Social reciprocity result from a fundamental inability to interpret and comprehend the feelings, intentions ,experiences and motives of others . The symptoms in this domain are as follows :

- children with ASD usually **have difficulties in interacting with their peers** due to impairments in the interpretation and use of social language including verbal and non verbal messages of others such as facial expressions , vocal inflections , gestures , social intentions and emotional tone .

- **Eye contact :** Typical people maintain eye contact with their conversational partners , whereas people with ASD will look at the person's mouth .

- Deficits in developing, maintaining, **and understanding relationships**, ranging, for example, from difficulties adjusting behavior to suit various social contexts; to difficulties in sharing imaginative play or in making friends; to absence of interest in peers.[4]

Although people with ASD have varying degrees of difficulty in initiating , responding and maintaining social interaction , they may be highly responsive to specific individuals or situations (e.g. , family members , favorite TV program) .Nonetheless , atypical patterns are noticed when relating to other people or strangers . [4]

Studies have shown that the primary neurological impairment in ASD is the dysfunctional mirror neuron system [4].Mirror neurons are brain cells in the premotor cortex which permits imitation of what the individual see , in addition they are involved in higher cognitive processes and language .Examples of

their function includes being able to imitate and learn from others' actions, or decode their intentions and empathize with their pain. In people with ASD , their mirror neurons respond only to what they do and not to the doings of others.[11]

2- Atypical communication development :

Language skills in children with ASD are extremely variable. Studies have shown that 9% of them remain nonverbal in later childhood . In general , the range of abilities extends from being non-verbal to verbose .[6]

Researchers classify Autism into two distinct phenotypes: **autism language normal** (ALN) in which language form is unimpaired and **autism language impaired** (ALI) in which language form is impaired in association with anomalies in left hemisphere structure and function .[6]

Language delay is the first area of concern identified by most families whose children are diagnosed later with ASD . Atypical language development may be related to comorbid intellectual disability or exist in isolation. In general , approximately 25-30% of children with ASD, early language development is typical but is subsequently lost between 18-24 months .[4]

Early language of children with ASD is often characterized by :

- **Imperative labeling** (using words for naming instead of communicating) , The use of language to share experiences and information , to inquire about other people ,to point out topics of interest is diminished .
- **Echolalia** : (echoing speech) , in normal development , echoing adult speech is common until the child understand words and learn to comprehend what is said to them . Echolalia diminishes before 2 years of age . However ,in ASD it may persist into childhood and beyond in the form of perseverative /uncontrollable stereotypes .
- Speech is characterized by , self-stimulation or singing .
- **Improper use of pronouns**, such as referring to self in the third person (e.g. saying “he wants banana vs I want Banana) .
- **Atypical prosody (inflection)** : Once functional language is established , prosody (use of pitch , loudness , tempo and rhythm in speech) maybe be robotic ,singsong or imitative in inflection.[4]

Children with ASD may have basic impairments in the in the use or comprehension of the nonverbal communication. Moreover, people with ASD may have a decreased ability to process speech and gestures simultaneously.

Studies have shown that brains of people with autistic disorder process faces as if they were objects , hence , every time the facial expression of the communication partner changes ,the person with ASD must re-identify the face . [4]

Children may lack the intention of communication and use their speech in idiosyncratic way ,e.g. the child may spell his mom's name rather than calling her mommy. In addition, They have very concrete and literal interpretation of words and barely use repair strategies when conversational breakdowns occur .[10]

In terms of cognition abilities , 50% to 70% of children with ASD obtain scores on nonverbal IQ measures of less than 70 .Executive functions tend to be impaired but working memory is an area of strength. In addition , the inability to recall word lists indicates impairments in the short term memory .[6]

3- Atypical behavior :

Restricted, repetitive patterns of behavior, interests, or activities are noted in the ASD . Below is a brief description of these symptoms as they appear in the DSM-V :

- **Stereotyped or repetitive** motor movements, use of objects, or speech (e.g., simple motor stereotypies, lining up toys or flipping objects, running in circles , hand waving , arm flapping ,toe walking echolalia, idiosyncratic phrases).

- **Insistence on sameness**, inflexible adherence to routines, or ritualized patterns or verbal nonverbal behavior (e.g., extreme distress at small changes, difficulties with transitions, rigid thinking patterns, greeting rituals, need to take same route or eat food every day). Interruptions of a ritual may upset the child with ASD and lead to distress and or a temper tantrum .

- Highly **restricted, fixated interests** that are abnormal in intensity or focus (e.g., strong attachment to or preoccupation with unusual objects, excessively circumscribed or perseverative interest).

- **Hyper- or hypo reactivity to sensory input** or unusual interests in sensory aspects of the environment (e.g., apparent indifference to pain/temperature, adverse response to specific

sounds or textures, excessive smelling or touching of objects, visual fascination with lights or movement) overreaction to certain environmental sounds (hyperacusis).

- **self-injurious behaviors .**

Associated conditions with autism spectrum disorders:

- **Intellectual disability** is commonly associated with ASD because the brain insults reposinle for causing ASD disrupt other neurologic functioning as well .
- **Learning disability** executive functions : the cognitive tast related to taking in , organizing , processing and asking on information is impaired in people with ASD and is sometimes manifested as ADHD (attention deficit hyperactivity disorder) .
- **Epilepsy** is present 25% of individuals with ASD .
- **Tic disorders** are brief involuntary movements are present in 9% of children with ASD .
- **Sleep disorders** are present in 50% - 70% of children with ASD including night wakening, delayed sleep onset and early morning waking.
- **Gastro intestinal** symptoms such as abdominal pain ,diarrhea ,malnutrition due to food selectivity and gastro esophageal reflux .

Early identification of autism spectrum disorders:

The age of identification of ASD depends on many factors including parent's and professional's knowledge of the child development .There appears to be a substantial gap between the time that parents first recognize a problem with their child development (onset of ASD) and the giving of a formal diagnosis . In most cases , parents often first become concerned with their child's development between 18 month and two years of age . The most common symptom to arouse concern is a delay of language acquisition.[5]

Early diagnosis is based on the recognition of the core symptoms in early childhood .Atypical development of pretend play , pointing to share interest ,use of eye gaze to engage another person in communication and social interest can distinguish children at risk for ASD as young as 18 months.[4]

Children with Asperger's disorder or high functioning autism tend to be diagnosed at school age ,when the social demands of the classroom make the symptoms more apparent as the requirements for social abilities increase with age .[5]

Usually , parents state that the child does not respond to his name and be less interested in faces and voices of others , has poor eye contact, absence of social mile, irritability and dislike of being hold. Those are the common signs of ASD **in infants** .[10] **In toddlers** ,sleep difficulties , limited diets , tantrums and inattention to language are the early signs reported by parents .

Common **Screening questions** for ASD :

- Does the child seem to want to communicate ?
- Is the child interested in playing or interacting with familiar adults or peers ?
- Does the child want to share his enjoyment with others ?
- Does the child respond to the attention of others ?
- Does the child engage in a variety of play activities or do things un a repetitive fashion ?
- Does the child play with toys in an appropriate or inappropriate fashion ? can he be easily distracted ?will the child point to desired objects , nod and shake his head in order to communicate ?[10]

Evaluation of autism spectrum disorders :

Multidisciplinary assessment is required for the evaluation and diagnosis of ASD . Diagnosis is made by a professional who is experienced in ASD such as a neurodevelopmental pediatrician ,developmental behavioral pediatrician ,child neurologist , child psychiatrist , child psychologist or speech language pathologist .[4]

Evaluation process requires obtaining information from several resources including :

- Interview with caregivers to obtain **case history** information about the early history of development , pregnancy and birth, associated medical problems and disorders , social development ,developmental millstones, especially in language ,family history including behavioral ,medical ,neurologic development and psychiatrist illness , current family functioning

and status . Case history information are used to verify whether the symptoms of ASD has been present in early infancy and childhood .

- **Observation** of the individual with ASD both in clinical settings and living environment.
- **Medical and neurological examination** aiming at ruling out hearing and vision loses ,any associated medical disorders , like tuberous sclerosis , the fragile x syndrome , other sensory deficits and other genetic disorders . Head circumference measures , skin examination are included within the medical .
- **Psychological evaluation** of the cognitive level of the individual with ASD. [5]
- **Language and developmental testing :**
    - Receptive and expressive vocabulary .
    - Assess response to name.
    - Nonverbal Cognitive abilities
    - Gross and fine motor function
    - Social and emotional skills.
    - Adaptive behavior .[6]

Intervention settings :

Assessment of the Baseline skills in socialization, communication and play is needed for the proper placement of the child in either of the following settings.

- Specialized setting :child receives highly intensive intervention (40hours/week) or less intensive (20hours/week) in a specialized segregated setting . Therapy is highly structured ,and focuses on strict behavioral management principles .
- Community settings : is the integration of children of ASD or PDD into community preschools /schools .The program should be flexible to accommodate the child's need , in order for the child to benefit from exposure and interaction with other adults and children .[10]

Treatment approaches:

ASD is one of the most variable disorders in terms of cognitive profile, language ability, comorbid diagnoses and eventual outcomes. Differences between children are striking as the similarities. E.g. an assessment or intervention approach that works well with one child may be completely inappropriate for another child with same ASD diagnosis.(6)

Intervention must be based on exposure, modeling, shaping and labeling done within a context of encouragement and structure to optimize the functional independence of the individual with ASD by minimizing the core symptoms of ASD ,to facilitate development and learning , to promote social skills of the individuals , to reduce the restricted interests and stereotypic behaviors and to eliminate maladaptive behavior.(10)

In this paper , the main focus is on two major areas of intervention which are behavior , speech and language therapy .

Behavior management approaches :

Key principle of behavioral management is not to extinguish the behaviors of ASD but to replace them with the more appropriate social , communicative and play skills . In this approach , the therapist creates a situation that precipitate target behaviors and then try to avoid or prevent that behavior or encourage the child to use a more appropriate one .(10)(4)

The most commonly used behavior management approaches with ASD are explained in details below :

1-Applied behavior analysis (ABA): is a system of autism therapy based on behaviorist theories which, simply put, state that behaviors can be taught through a system of rewards and consequences. The first step in ABA is to analyze the behavior and this is done using the **ABC model:**

A - Antecedent
A directive or request for the child to perform an action.
B - Behavior
A behavior, or response from the child - successful performance, noncompliance, or no response.
C - Consequence
A consequence, defined as the reaction from the therapist, which can range from strong positive reinforcement .

ABA employs **strategies** based on scientific principles of behavior that are designed to build socially useful repertoires and reduce problematic ones. These include :

Task Analysis : is a process in which a task is analyzed into its components so that those parts can be taught through the use of chaining: forward chaining, backward chaining and total task presentation.

Chaining : is breaking down /segmenting of a skill into the smallest units for easy learning.

Prompting

The parent or therapist provides assistance to encourage the desired response from the child. The aim is to use the least intrusive prompt possible that will still lead to the desired response. Prompts can include: Verbal cues , Visual cues , Physical guidance and Demonstration .

Fading : is the gradual elimination of prompts as the new behavior is learned , so the child does not become overly dependent on them when learning a new behavior or skill.

Shaping : Shaping involves gradually modifying the existing behavior of a child into the desired behavior. This is paired with positive .

Differential reinforcement : Reinforcement is a consequence of a child's behavior that will most likely increase that behavior. It is "differential" because the level of reinforcement varies depending on the child's response. Reinforcement can be positive (verbal praise or a favorite activity) or negative (an emphatic 'no').

Generalization : Once a skill is learned in a controlled environment the skill is taught in more general settings. Perhaps the skill will be taught in the natural environment.

Video modeling : One teaching technique found to be effective with children, is the use of video taped sequences as exemplars of behavior. It can be used by therapists to assist in the acquisition of both verbal and motor responses, in some cases for long chains of behavior.

Discrete trial training (DTT) : is a particular ABA teaching strategy which enables the learner to acquire complex skills and behaviors by first mastering the subcomponents of the targeted skill. By utilizing teaching techniques based on the principles of behavior analysis, the learner is gradually able to complete all subcomponent skills independently. Once the individual components are acquired, they are

linked together to enable mastery of the targeted complex and functional skill. This methodology is highly effective in teaching basic communication, play, motor, and daily living skills.
Specifically, there are a number of **weaknesses** with DTT including that it is primarily teacher initiated, that typically the reinforces used to increase appropriate behavior are unrelated to the target response, and that rote responding can often occur.

A discrete trial consists of four parts:

• **An instruction** stated as concisely as possible, for example, "Come here," versus "I want you to come here, please."

• **A response** from the child that may take several forms including successful completion ,partial success, noncompliance, or unresponsiveness.
The latter two would be considered incorrect responses.
• A **consequence** provided by the practitioner that corresponds to the child's response..
• **An inter-trial interval**. This refers to the teacher pausing briefly after providing a consequence and before starting the next trial. An inter-trial interval helps to ensure that each trial is discrete from the next trial.[12]

Natural Environment Training (NET) may be used to address more complex skills. NET specifically addresses the above mentioned weaknesses of DTT in that all skills are taught in a more natural environment in a more "playful manner." Moreover, the reinforcers used to increase appropriate responding are always directly related to the task (e.g., a child is taught to say the word for a preferred item such as a "car" and as a reinforcer is given access to the car contingent on making the correct response).

3-Developmental Individual-difference Relationship-based model (DIR) is a comprehensive, interdisciplinary approach developed by Drs. Stanley Greenspan and Serena Wieder. The approach focuses on using an intensive integrated intervention that aims to help children expand their circles of communication by meeting them at their developmental level and building on their strengths. The ultimate goal of the intervention is to help the child develop appropriate affect and form a sense of self as an intentional and interactive individual. Parents and primary caregivers are taught specific strategies which are utilized during eight to ten sessions (20-30 minutes each) per day of "floor time" play with the child.

"Floor time" is a series of reciprocal child-directed interactions resulting in "communication circles." The goal of "floor time" is to sustain interactions between the child and the adult by gradually increasing the circles of communication. These interactions become the basis for further play development.

During "floor time," the adult joins in the child's play to increase opportunities for pleasurable interaction and engagement. The child's actions are considered intentional and purposeful. The adult follows the child's lead to validate the child's sense of self. Preferred objects and activities are used to motivate the child and facilitate persistence and patience. Higher level skills and concepts are taught through interactive play. "Floor time" also provides for peer experiences with typically-developing age-mates.[12]

4-Relationship Development Intervention

RDI tries to help children interact positively with other people, even without language. When children learn the value and joy of personal relationships, according to RDI, they will find it easier to learn language and social skills. RDI is based on the idea that children with autism missed some or many of the typical social development milestones as infants and toddlers. They can be given a "second chance" to learn these skills through play, "guided participation" and other activities, according to RDI.

Here's an example of RDI in action: "The adult holds a treat in one closed fist, displays both closed fists to the child, and then looks at the hand that holds the treat. The child is given repeated opportunities to 'find' the treat in the hand the adult looks at," .

Speech language therapy :

The goal of speech and language intervention is to encourage the social use of language , to develop more age-appropriate communication skills , to expand the repertoire of communicative functions and develop imitation and play skills .Any form of communication should be encouraged , whether it involved speaking , pointing , gestures or any other form of non-verbal communication . Thus , it is important to recognize the preliminary attempts to communicate and respond to them .

For children with no speech who are showing communicative intent , signs and other alternative augmentative communication (ACC) may be presented as a bridge to speech , but vocal imitation and speech production should be a focus of intervention for these children . [6]

Working on Receptive language : Studies by Adamson and colleagues , indicate the symbol infused joint attention , that is , the amount of time spent attention to a shared object that the communication partner is naming and talking about , is related to growth of receptive as well as expressive language .

Working on receptive language will include :

- Proving highly engaging joint attentional opportunities, by sharing interesting objects and activities with the client.
- Actively attracting their attention and gaze to the object and to the communication partner .
- Providing simple , repetitive language to accompany the activity .
- Preissler (2008) , found that when taught names of pictures , children with ASD did not realize the real objects the pictures represented . this suggests that children ASD have difficulty understanfing that pictures are representations of objects .thus , it is important to teach language in relation to real , functional objects rather than pictures with this population . [6]

Working on vocabulary and utterance length :

techniques used **in milieu teaching** such as following the child's attentional lead ,labeling objects the child shows interest in , withholding those objects while looking expectantly at the child . . All aides in the increase of expressive vocabulary in children with ASD . [6]

Script therapy : Scripts are verbal statements in either written or in an audio format. An individual is taught to repeat the script in appropriate specific social situations (e.g., "At the park I play on the slide."). As individuals learn to use the scripts, they are faded, typically one word at a time, from end to beginning (e.g., "At the park I play on the ____").[6]

Working with Echolalia:

our work with children who display immediate or delayed echolalia should focus on enhancing the attention to speech and receptive language ability , increasing spontaneous spoken vocabulary and utterance length and to work with echolalia to increase its functionality. [6

: Approaches used to decrease echolalia include

- **using a third person** (or puppet) to model what should be said . this game can be turned into a turn taking game , in which the puppet makes a statement and the child imitates it in order to accomplish what the puppet did .

- **mitigated echolalia** : when an echoed utterance is produces , the clinician can echo it back, then add a slight change and invite imitation of the change .Once this can be done in short phrases , longer utterances can be used .

Concluding comments (FACTS ABOUT ASD)

1 . Autism Is a 'Spectrum' Disorder

2 . Asperger Syndrome is a High Functioning Form of Autism

3 . People With Autism Are Different from One Another

4 . There Are Dozens of Treatments for Autism - But No 'Cure'

5 . There Are Many Theories on the Cause of Autism, But No Consensus

6 . Children Rarely "Outgrow" or "Overcome" Autism

7 . Families Coping with Autism Need Help and Support

8 . There's No 'Best School' for a Child with Autism

9 . There Are Many Unfounded Myths About Autism

10 . Autistic People Have Many Strengths and Abilities

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Rett Syndrome

Introduction

Rett syndrome is a disorder that was first identified by Dr. Andreas Rett, an Austrian physician who first described it in a journal article in 1966. It was not until after a second article about the disorder, published in 1983 by Swedish researcher Dr. Bengt Hagberg, that the disorder was generally recognized. (2)

Definition

Rett syndrome is a neurodevelopmental disorder that is classified as pervasive developmental disorder . It is a rare, genetic disorder which affects the development of the brain.. (2)**.** It is most often being misdiagnosed as autism or cerebral palsy. (1)

Causes of Rett syndrome:

The majority of the cases are caused by mutation of the MECP2 gene which in involved in producing a protein that is essential for brain development. The gene appears on the X chromosome

Not everyone who has an *MECP2* mutation has Rett syndrome. Scientists have identified mutations in the *CDKL5* and*FOXG1* genes in individuals who have atypical or congenital Rett syndrome, but they are still learning how those mutations cause the disorder. Scientists believe the remaining cases may be caused by partial gene deletions, mutations in other parts of the *MECP2* gene, or additional genes that have not yet been identified, and they continue to look for other causes. (4)

Seventy to 80 percent of girls given a diagnosis of Rett syndrome have the MECP2 genetic mutation detected by current diagnostic techniques. Scientists believe the remaining 20 to 30 percent of cases may be caused by partial gene deletions, by mutations in other parts of the gene, or by genes that have not yet been identified; thus, they continue to search for other mutations. (2)

Is Rett syndrome inherited?

Although Rett syndrome is a genetic disorder, less than 1 percent of recorded cases are inherited or passe d from onegeneration to the next. Most cases are spontaneous, which means the mutation occurs randoml y. However, in somefamilies of individuals affected by Rett syndrome, there are other female family me mbers who have a mutation of their*MECP2* gene but do not show clinical symptoms. These females are k nown as "asymptomatic female carriers." (2)

Who get Rett syndrome?

Rett syndrome is estimated to affect one in every 10,000 to 15,000 live female births (1)

- **Boys**

  Rett Syndrome in boys: Because boys have one X chromosome and one Y chromosome, the X chromosome can't be inactivated. Therefore, boys can't be protected from the severe effects of the disease. They usually die at birth or a few months afterward .

- **Girls**

  Rett syndrome in girls: Because girls have two x chromosomes and a cell only needs one copy of the gene that works, each cell inactivates one of its chromosomes. The more cells that have the gene inactivated, the less severe the symptoms. (3)

The course of Rett syndrome

The course of Rett syndrome, including the age of onset and the severity of symptoms, varies from child to child. Before the symptoms begin, however, the child generally appears to grow and develop normally, although there are often subtle abnormalities even in early infancy, such as loss of muscle tone (hypotonia), difficulty feeding, and jerkiness in limb movements. Then, gradually, mental and physical

symptoms appear. As the syndrome progresses, the child loses purposeful use of her hands and the ability to speak. Other early symptoms may include problems crawling or walking and diminished eye contact. The loss of functional use of the hands is followed by compulsive hand movements such as wringing and washing. The onset of this period of regression is sometimes sudden. (2)

Most people with Rett syndrome live to be between 40-50 and need continuous care throughout their life. (1)

Rett syndrome signs and symptoms include:

- **Slowed growth.** Brain growth slows after birth. Smaller than normal head size is usually the first sign that a child has Rett syndrome. It generally starts to become apparent after 6 months of age. As children get older, delayed growth in other parts of the body becomes evident.
- **Loss of normal movement and coordination.** The most significant loss of movement skills (motor skills) usually starts between 12 and 18 months of age. The first signs often include a decrease of hand control and a decreasing ability to crawl or walk normally. At first, this loss of abilities occurs rapidly and then continues more gradually.
- **Loss of communication and thinking abilities.** Children with Rett syndrome typically begin to lose the ability to speak and to communicate in other ways. They may become uninterested in other people, toys and their surroundings. Some children have rapid changes, such as a sudden loss of speech. Over time, most children gradually regain eye contact and develop nonverbal communication skills.
- **Abnormal hand movements.** As the disease progresses, children with Rett syndrome typically develop their own particular hand patterns, which may include hand wringing, squeezing, clapping, tapping or rubbing.
- **Unusual eye movements.** Children with Rett syndrome tend to have unusual eye movements, such as intense staring, blinking or closing one eye at a time.
- **Breathing problems.** These include breath-holding (apnea), abnormally rapid breathing (hyperventilation), and forceful exhalation of air or saliva. These problems tend to occur during waking hours, but not during sleep.

- **Irritability.** Children with Rett syndrome become increasingly agitated and irritable as they get older. Periods of crying or screaming may begin suddenly and last for hours.
- **Abnormal behaviors.** These may include sudden, odd facial expressions and long bouts of laughter, screaming that occurs for no apparent reason, hand licking, and grasping of hair or clothing.
- **Seizures.** Most people who have Rett syndrome experience seizures at some time during their lives. Symptoms vary from person to person, and they can range from periodic muscle spasms to full-blown epilepsy.
- **Abnormal curvature of the spine (scoliosis).** Scoliosis is common with Rett syndrome. It typically begins between 8 and 11 years of age.
- **Irregular heartbeat (dysrhythmia).** This is a life-threatening problem for many children and adults with Rett syndrome.
- **Constipation.** This is a common problem in people with Rett syndrome. [3]

Development of Rett syndrome

The developmental stages of Rett syndrome are as follows:

- **Stage 1: Early Onset (6-18 months)**

Slowing of an infant's head growth after 5 months, may not make eye contact, and may not show interest in toys. Could be calm, quiet, and use repetitive hand movements such as hand washing or clapping. Most girls crawl without using their hands. This stage usually lasts for a few months but can continue for more than a year.

- **Stage 2: Rapid Destructive (1-4 yrs. old)**

Severe impairments in speech and the lack of ability to perform motor functions, including chewing and swallowing. Repeated hand stereotypes become common along with possible abnormal sleeping patterns, teeth grinding, and loss of muscle tone. Evident incidents of breath holding or hyperventilating can occur. May become irritable because of communication barriers and walk unsteadily. Slowed head growth is usually noticed during this stage.

- **Stage 3: Plateau (2-10 yrs. old)**

Motor development is delayed (Apraxia), gastrointestinal disorders and seizures often appear. The child's behavior frequently shows some improvement such as less irritability and crying, better

communication skills, and an increase in attention span. Many patients diagnosed with RS stay in this stage for most of their lives.

- **Stage 4: Late Motor Deterioration (Usually after age 10)**

Can last for years or decades. Prominent features include reduced mobility, curvature of the spine (scoliosis) and muscle weakness, rigidity, spasticity, and increased muscle tone with abnormal posturing of an arm, leg, or top part of the body. Girls who were previously able to walk may stop walking. Cognition, communication, or hand skills generally do not decline in stage IV. Repetitive hand movements may decrease and eye gaze usually improves. [(2)]

Diagnosis of Rett syndrome

A pediatric neurologist, clinical geneticist, or developmental pediatrician should be consulted to confirm the clinical diagnosis of Rett syndrome. Many early cases of RTT are initially misdiagnosed as autism, especially when the signs of the disorder first become apparent. Until recently RTT was considered to be part of the autism spectrum, but with the release of the new DSM 5 this year, it is no longer listed as an autism spectrum disorder. [(4)]

The physician will use a highly specific set of guidelines that are divided into three types of clinical criteria: ***main***, ***supportive***, and ***exclusion***. The presence of any of the exclusion criteria negates a diagnosis of classic Rett syndrome.

Main criteria:

- Partial or complete loss of functional hand skills
- Partial or complete loss of spoken language skills
- Impaired apraxic gait or absence of ability to ambulate
- Stereotypic, repetitive, nonfunctional hand movements

Exclusion Criteria:

- A diagnosis of RTT cannot be made if there is:
- Any acquired injury to the brain, neurometabolic disease, or infection resulting in neurological impairment
- A history of abnormal psychomotor development in the first six months of life

Supportive Criteria - often observed, not required

- Respiratory disturbances, such as rapid breathing or breath holding
- Teeth grinding
- Impaired sleep patterns: frequent sleeping during the day, frequent night waking
- Abnormal muscle tone
- Peripheral vasomotor disturbances
- Small cold hands and feet
- Scoliosis and/or kyphosis
- Growth retardation
- Laughing and/or screaming spells, not appropriate for the context
- Lessened response to pain
- Intense eye communication – often referred to as "eye pointing" [5]
-

What treatment is available?

There is no cure for Rett syndrome. Treatment for the disorder is symptomatic — focusing on the management of symptoms — and supportive, requiring a multidisciplinary approach. Medication may be needed for breathing irregularities and motor difficulties, and antiepileptic drugs may be used to control seizures. There should be regular monitoring for scoliosis and possible heart abnormalities. Occupational therapy (in which therapists help children develop skills needed for performing self-directed activities such as dressing, feeding, and practicing arts and crafts), physiotherapy, and hydrotherapy may prolong mobility. Some children may require special equipment and aids such as braces to arrest scoliosis, splints to modify hand movements, and nutritional programs to help them maintain adequate weight. Special academic, social, vocational, and support services may also be required in some cases. [6]

Swallowing Deficits

80% of individuals with Rett syndrome have reported swallowing deficits [7]

Risks:

- Malnutrition and growth failure
- Potential for aspiration [8]

Oropharyngeal dysfunction

- Inadequate lip closure
- Impaired chewing
- Poor tongue mobility-weak base of tongue retraction
- Reduced oral and pharyngeal transit time and clearance
- Penetration of liquids and solids during the swallow
- Many studies report no overt aspiration

Gastroesophageal dysfunction

- Esophageal dysmotility
- Delayed emptying
- Spasms
- Gastroesophageal reflux [7] [8]

Feeding/Swallowing Recommendations

Swallowing/Feeding evaluation

- Nutritional assessment
- Early analysis of texture tolerance

- Diet modification- less chewy textures [9]

Feeding

- Maintaining self-feeding- hand on utensil may give feedback to coordinate breathing/feeding pattern
- Feeder Training- needs to be aware of breathing patterns potential breath holding[8]

Feeding Aids

- specialty cups and bottles; adjusted seating/specialty chairs[10]

Survey of communication in RTT

- We all communicate using multiple modalities – individuals with RTT do as well
- They use eye gaze most frequently, followed by picture/symbol boards (accessed by switches or hands) and body movement
- Exposure to AAC should begin EARLY
- Educators should evaluate all modalities (gaze, gestures, vocalizations, pictures/symbol use)
- Not all modalities work at all times
- Consider:

influence of apraxia, delayed response time, inconsistency, level of awareness

<u>Communication intervention needs to:</u>

- begin early
- be modified as the individual with RTT grows and changes
- Include all stakeholders
- offer multiple options for communication to meet the needs of

- different situations [5]

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Childhood disintegrative disorder

Definition

Childhood disintegrative disorder (CDD) is also known as Heller's syndrome. It's a very rare condition in which children develop normally until at least two years of age, but then demonstrate a severe loss of social, communication and other skills.

Childhood disintegrative disorder is unlike autism, someone with childhood disintegrative disorder shows severe regression after several years of normal development and a more dramatic loss of skills than a child with autism does. In addition, childhood disintegrative disorder can develop later than autism does.

Causes of CDD

The cause of childhood disintegrative disorder is unknown. Research findings suggest, however, that it may arise in the neurobiology of the **brain** . About half the children diagnosed with CDD have an abnormal electroencephalogram (EEG). EEGs measure the electrical activity in the brain generated by nerve transmission (brain waves). CDD is also sometimes associated with **seizures**, another indication that the neurobiology of the brain may be involved. CDD is occasionally associated with such diagnosed medical disorders of the brain as Schilder's disease; but no one disease, brain defect, disorder, or condition can account for all symptoms and all cases. Research is hampered by the rarity of this disorder. Occurs more in males than females.

Symptoms of CDD

Children with CDD have at least two years of normal development in all areas language understanding, speech, skill in the use of large and small muscles, and social development. After this period of normal growth, the child begins to lose the skills he or she has acquired. This loss usually takes place between ages three and four, but it can happen any time up to age ten.

The loss of skills may be gradual, but more often occurs rapidly over a period of six to nine months. The transition may begin with unexplained changes in behavior, such as anxiety, unprovoked anger, or agitation. Behavioral changes are followed by loss of communication, social, and motor skills. Children may stop speaking or revert to single words. They often lose bowel or bladder control and withdraw into themselves, rejecting social interaction with adults or other children. They may perform repetitious activities and often have trouble moving from one activity to the next.

Diagnosis of CDD

For a child to be diagnosed with childhood disintegrative disorder, he or she must meet the criteria in the Diagnostic and Statistical Manual of Mental Disorders (DSM) published by the American Psychiatric Association, summarized below:

Normal development for at least the first 2 years of life

Normal development includes age-appropriate verbal and nonverbal communication, social relationships, and motor, play and self-care skills.

Significant loss of previous skills

Loss of previous skills, after at least two years of normal development, in at least two of the following areas:

- Ability to say words or sentences (expressive language)
- Ability to understand verbal and nonverbal communication (receptive language)
- Social skills and self-care skills (adaptive behavior)
- Bowel and bladder control
- Play skills
- Motor skills (ability to voluntarily move the body in a purposeful way).
-

Lack or loss of normal function

Lack or loss of normal function occurs in at least two of the following areas:

- **Social interaction.** This may include a wide range of problems with social connectedness. Your child may have difficulty with nonverbal interactions, may not make friends with peers, and may lack the ability to share, recognize, understand and respond to others' social cues and feelings.
- **Communication.** This may include a delay or loss in the ability to speak or to start and maintain conversations. Your child also may use the same words over and over, and may not "get" imaginative or make-believe play.
- **Repetitive and stereotyped patterns of behavior, interests and activities.** Your child may flap his or her hands, rock or spin (motor stereotypes and mannerisms); may become attached to specific routines and rituals; or may have difficulty with transitions or changes in routine. Many children with the disorder develop a fixed posture or body position (catatonia) and may become preoccupied with certain objects or activities.

Screening

Your child's doctor should perform developmental screenings at well-child visits or if you suspect that there's a delay in your child's development or a loss of age-appropriate skills.

If your doctor sees signs or symptoms of a developmental disorder or delay, your child may be referred to one or more specialists for evaluation and diagnosis. These may include a child psychologist, a child psychiatrist, a doctor who specializes in conditions of the brain and nervous system (neurologist), a pediatrician specializing in behavioral and developmental problems, a hearing specialist (audiologist), a speech therapist, a physical therapist, and an occupational therapist.

These professionals may perform some or all of the following tests:

- **Medical history.** This is an extensive interview with special emphasis on when developmental milestones were reached and the age at which previously learned skills were lost. Baby books, family photo albums and videotapes may help you accurately remember when your child reached specific developmental milestones.
- **Neurological exam.** A neurologist performs a physical exam to look for abnormalities in your child's brain and nervous system. Your neurologist may order imaging tests of the brain and tests that measure the brain's electrical activity.
- **Genetic tests.** These tests usually involve a blood test to study your child's chromosomes and determine if there's an inherited family condition or disease.
- **Communication and language tests.** In-depth tests can measure how your child communicates with words and nonverbal gestures (facial expressions, posture, rhythm of speech, gestures) and how your child interacts with others (understanding words, body language, social cues, tone of voice).
- **Lead screening.** A blood test is performed to check for lead poisoning. Lead exposure in children causes damage to the nervous system, developmental delays, hearing loss and behavior problems.
- **Hearing (audiology) test.** This is an exam to check for hearing loss or hearing-related problems.
- **Vision test.** This exam checks for vision loss or vision-related problems.
- **Behavior inventory.** Doctors use formal rating scales and checklists to document the occurrence of specific behaviors, such as repetitive movements, oversensitive or undersensitive responses to normal sights, sounds and touch sensations in the environment, as well as social interactions and play skills.

Developmental tests

Your doctor may also want your child to have several developmental tests to measure how your child performs skills compared with other children of the same age. These tests measure the following skills:

- **Large motor skills.** This includes walking, running, jumping, throwing and climbing.
- **Fine motor skills.** This is the use of hands and fingers for the manipulation of small objects, such as buttons, pencils and scissors.
- **Sensory skills.** This is how the brain and body organize and respond to a variety of everyday sounds, sights, smells, tastes and touch (tactile) experiences in the environment.
- **Play skills.** This involves how your child plays with toys and other objects as well as with children and adults. The style and type of play behavior (imaginative, varied, purposeful, goal-directed) are observed.
- **Self-care skills.** These skills include toileting, feeding, dressing and brushing teeth.
- **Cognitive skills.** These skills include the ability to pay attention, follow directions, think, concentrate and solve problems.

Test results allow your health care team to look for underlying medical or neurological conditions that may be causing child's signs and symptoms, rule out other conditions or diseases, and make an accurate diagnosis. Then the team can develop the best treatment plan for your child

Treatment of CDD

There's **no cure** for childhood disintegrative disorder. Treatment for the disorder is basically the same as for autism.

Treatments to relieve or lessen symptoms may include:

- **Medications.** There are no medications that directly treat childhood disintegrative disorder. However, severe behavior concerns that can threaten safety, such as excessive impulsiveness or repetitive movements, may sometimes be controlled by medications for anxiety or depression, or antipsychotic medications. Anticonvulsant drugs may help control epileptic seizures.
- **Behavior therapy.** This therapy technique may be used by psychologists, speech therapists, physical therapists and occupational therapists, as well as parents, teachers and caregivers. Behavior therapy programs may be designed to help your child relearn or minimize the loss of language, social and self-care skills. These programs use a system of rewards to reinforce desirable behaviors and discourage problem behavior. A consistent approach among all health care team members, caregivers and teachers is important in behavior therapy.

Although abilities and behaviors vary greatly for children with childhood disintegrative disorder, the outcome is worse than for children with autism. The loss of language, cognitive, social and self-care skills

tends to be severe and unlikely to improve. Children with the disorder generally need lifelong support with the activities of daily living, and may eventually need residential care in a group home or long-term care facility.

Prognosis of CDD

The prognosis for children with CDD is very poor; it is worse than the prognosis for children with autism. Once skills are lost, they are not usually regained. Only about 20% of children diagnosed with the disorder reacquire the ability to speak in sentences. Most adults with CDD remain dependent on full-time caregivers or are institutionalized.

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ORGANIZATIONS

Autism Society of America. 7910 Woodmont Avenue, Suite 300, Bethesda, MD 20814-3067. (301) 657-0881 or 800-3AUTISM.

National Association of Rare Disorders (NORD). P.O. Box 8923, New Fairfield, CT 06812-8923. (800) 999-NORD or (203) 746-6518.

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