Imagine the symptoms of autism, cerebral palsy, Parkinson’s, epilepsy, and anxiety disorders—all in one little child.

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Publications
GOAL FOR TODAY

- Overview of Rett Syndrome characteristics & Recent updates on Genetic Findings
- What to expect from AAC users who have Rett Syndrome
- AAC system recommendations for Individuals with Rett Syndrome

HISTORY OF RETT SYNDROME

1954 Dr. Rett first noticed patients sharing similar symptoms. He published his first findings in 1966 in German and in 1977 in English.

1983 an article on Rett syndrome appeared in the mainstream, English-language journal, Annals of Neurology. Written by Dr. Hagberg and his colleagues, the report finally raised the profile of Rett syndrome and put it on the radar screens of many more investigators. This article was a breakthrough in communicating details of the disease to a wide audience, and the authors honored its pioneering researcher by naming it Rett syndrome.

A major breakthrough occurred in 1999, when a research fellow at Baylor named Ruthie Amir discovered MECP2, the gene that, when mutated, causes Rett syndrome. The discovery of the gene, located at the Xq28 site on the X chromosome.

WHAT IS RETT SYNDROME (RTT)

Neurodevelopmental genetic disorder resulting in:
- loss of functional movement
- loss of speech
- loss of swallowing, feeding

- Not degenerative—live well into adulthood
- Complex and unexplained cognitive and behavioral issues of normal development
- Condition of developmental arrest
- Not hereditary: spontaneous mutation (Mutation of the methyl-CpG binding protein-2, MECP2)
- Range of abilities/disabilities (typical vs. atypical)
GENETIC EXPLANATION

MECP2 on the X chromosome
Plays a role in brain development
Encodes one protein responsible for directing other genes to turn on/tum off
Disrupts the regulated pattern of development

AMIR & ZOGHBI, 2000

DIAGNOSTIC CRITERIA

Neul et al. described revised diagnostic criteria. Classic RTT requires apparently normal psychomotor development in the first 6 months of life followed by a period of regression, which is not due to brain injury secondary to trauma, neuroendocrinologic disease or severe infection, and involves partial or complete loss of acquired purposeful hand skills and language, gait abnormalities and the development of stereotypic hand movements, followed by stabilization or even some degree of recovery. An important aspect of the regression is a period of social withdrawal or impaired communication.

Atypical RTT requires a similar period of regression and subsequent stabilization/recovery, at least two of the above four behavioral manifestations and the presence of at least five (out of 11) supportive criteria.


CLASSIC 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
- Irregular breathing patterns, breath holding or hyperventilation (only when awake)
PREVALENCE

As prevalent as Cystic Fibrosis, ALS and Huntington’s, another little girl is born with Rett Syndrome every 90 minutes.

1:10,000

MALES WITH RETT SYNDROME

Because MECP2 is on an X-linked gene, it had been thought that Rett syndrome only affected females. However, we now know that males born with a mutation in MECP2 may also be affected.

3 possible genetic mutations leading to Rett syndrome may occur in males:

1. A male with Klinefelter Syndrome is born with an extra X chromosome (XXY). If one X chromosome has the MECP2 mutation, these males will exhibit Rett symptoms similar to females.
2. A MECP2 mutation may occur sporadically later in human development. In the case of a 2 chromosome, it breaks the mutation. Rather, only one of the MECP2 mutations occurs in males, whereas with females, the mutation will affect both the X chromosome and the Y chromosome, leading to Rett syndrome.
3. A MECP2 mutation may occur sporadically later in human development. If one X chromosome has the MECP2 mutation, the mutation is randomly inactivated. Therefore, a male with this type of mutation will exhibit Rett syndrome similar to females.

Males exhibiting the typical sex chromosomes (XY) but have a MECP2 mutation exhibit milder forms of Rett syndrome in which the X chromosome is randomly inactivated. However, they do not exhibit symptoms as severe as females. The symptoms are less severe because, in males, they have one normal X chromosome to compensate for the mutation.

It is thought that around 1 in 10,000 males have Klinefelter Syndrome, whereas there are no known cases of males with Rett Syndrome caused by sporadic mutations, which are likely to occur at a much lower frequency. However, it is possible that some males with Rett Syndrome may have this genetic defect, but their symptoms may not manifest in the same way as females.
CHARACTERISTICS

- Hand Stereotypies
  - Wringing, picking, mouthing, hands to hair
- Breathing Abnormalities
  - Breath holding, breathing rapidly
- Anxiety and Fear
- Disturbance in Sleep Patterns (80%)  
  - Nighttime laughing, daytime sleepiness, problems staying asleep & problems getting to sleep, sleep disordered breathing (sleep apnea)
- Cognitive Impairments (severe)
  - Motor function (walking, swallowing, hand movements)

These other characteristics are less prevalent but do appear in the population

- Screaming
- Teeth grinding
- Self-injurious behavior

Although most purposeful movements across the body are impacted, eye-gaze remains intact leaving it a strong potential access point for communication.

SEVERE APRAXIA

Apraxia (dyspraxia), the inability (or reduced ability) to program the body to perform motor movements, is the most fundamental and severely handicapping aspect of RTT.

Accepted explanations of why the girls fail to regain speech

- Apraxia impacts ALL motor movements

Apraxia symptoms increase when....

  - feel rushed
  - feel pressure
  - demands are placed
CHARACTERISTICS

Pre-intentional
- Caregivers assign meaning to an individual's behavior and children have no expectation or awareness that caregivers will respond.
- Behaviors are only considered communicative because their partners interpret them as intentional.
- At risk for failure
- Hard to include them in meaningful ways

LEVELS OF COMMUNICATION

Pre-intentional
- Caregivers assign meaning to an individual's behavior and children have no expectation or awareness that caregivers will respond.
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MOTHERS INTUITION

NOT SURE IF I SHOULD TAKE A MAP
OR CRY ABOUT BEING TIRED

LEVELS OF COMMUNICATION

Emerging Intentional: Pointing to objects and people, looking at things and reaching.
At this stage, the child's behavior is now intentional and directed to another person, but not yet symbolic. They can make someone in their environment do something, but do not express this desire through conventional communication (e.g., spoken words).

Is a behavior, like vocalizing or moving a body part, purposeful?
1. Does the client make eye contact when demonstrating that behavior?
2. Is it something he/she seems to do consistently in certain circumstances?
3. Does he/she alternate looking between the communication partner and the object?
4. Is he/she persistent and try again if the initial attempts are not satisfied?
5. Does he/she seem to modify the behavior in an attempt to be recognized/noticed?

Preintentional Perlocutionary Stage
Intentional, but PreSymbolic Illocutionary State
Locutionary Stage

Intentional Stage
Individual communicates an intended message via speech, gestures, behavior or symbolically in the same way repetitively.

Symbolic Stage
In typical children, this stage begins with first words.
For those who cannot communicate through natural speech, conventional signs language or intentional use of AAC symbols (adapted or pictures).
Follows modeling from communication partners.

LEVELS OF COMMUNICATION
IS IT INTENTIONAL?

If you're unsure, treat the behavior AS IF it is a true communication attempt. Doing so increases the chances that the learner will keep at it. And that, of course, is exactly what we want.

COMMUNICATION ASSESSMENT

The Inventory of Potential Communicative Acts (IPCA) (Sigafos et al, 2006)

Each of these tools use a series of questions to gather information from a communication partner of the individual you are assessing. These are highly functional ways to gather useful information about communication and social behaviors of individuals who are at a pre-linguistic or early-linguistic level of communication development.

TRAINING MATERIALS FOR TEACHERS

http://complexneeds.org.uk/
Training for teachers of learners with severe, profound and complex learning difficulties.
Module 2.2 Considering communication and interaction
Module 2.4 Assessment, monitoring and evaluation
Module 3.1 Communication-augmentative and assistive strategies
ASSUME COMPETENCE

Many people (parents, caregivers and professionals), believe that the girls are “locked in,” and unable to demonstrate what they know and understand. This is important to acknowledge, even as a remote possibility, because what we believe about an individual affects the way we interact with them.

STRATEGIES

1. Attribute meaning to communication attempts even if meaning is uncertain
   In at least one study, symbolic communication seemed to increase as a function of improving mothers’ awareness of and ability to acknowledge the many communication modes used by their daughters.
2. Prompt use of communication system through natural means rather than commands
3. Provide sufficient wait time and a hierarchy of support
4. Consistently ask questions and make comments that maximize use of available symbols and voice output messages as appropriate communication turns.


BEST PRACTICES IN AAC INTERVENTION FOR RTT

- Teach requesting
- Give choices
- Develop consistency
- Expand to 3 or more choices
- Allow for inconsistency because of apraxia

SENSORY REGULATION

The Inverted-U Model
Balancing Pressure and Performance
Also known as Yerkes-Dodson Law

FLOW

SENSORY REGULATION

Calming Activities
- Repetitive predictable movements
- Deep sustained touch
- Deep pressure
- Linear vestibular input
- Oral motor activities
- Rhythmic low-intensity auditory input
- Low intensity slightly similar visual input

Alerting Activities
- Quick unexpected movements
- Speed changes, direction changes
- Rotary movement
- Loud noises, music
- Light touch
- High contrast visual input
- Strong intensity taste or smell
**TIPS FOR CHOOSING TASKS**

- Set clear goals which are understood, attainable and within person’s abilities.
- High degree of concentration on limited field.
- Immediate feedback.
- Neither too easy nor too hard.
- Sense of personal control.
- Change activities if individual is no longer “in the zone.”

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**THINK OF THE LOAD**

![Diagram showing Cognitive and Motor loads](image)

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**VOCABULARY**

- Vocabulary should be words that can be used in many settings (Core Vocabulary).
- Vocabulary can also be highly motivating items/actions.
- Vocabulary should always be positioned consistently in the same location.

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COMMUNICATION COACHING

- Observe communication partner interact with individual with RTT
- Help partners identify behaviors which may be intentional or communicative in nature
- Provide strategies to enhance these behaviors during familiar routines
- Provide feedback
- Model behaviors
  - Increasing wait time
  - Partner aided scanning
  - Giving choices

A CONVERSATION WITH MAGGIE

https://www.youtube.com/watch?v=R_1U4JmaYRR

EYE-GAZE TECHNOLOGY

- Suggestions for implementation
  - First establish consistent eye gaze using high-interest objects, photos or symbols
  - It can be very challenging to achieve calibration for individuals with RTT – go slow, and don’t be discouraged. It takes time to learn how to activate buttons using eye gaze
  - Begin slowly using high-interest activities (Look to Learn software)
  - Trial a device before recommending a purchase (as you would with other clients)
  - Remember that one system won’t work in all contexts, so explore multiple communication options
PRC ACCENT WITH NUEYE

TOBII DYNAVOX

LOOK TO LEARN

Five Areas of Learning
The 40 activities are split over five key areas.
Sensory – Designed to teach cause and effect
Explore – Encourages the user to engage with the whole screen
Target – Helps improve accuracy of eye gaze access
Choose – Develops choice making skills
Control – Fine tunes eye gaze access

*Free trial version
ANNA

Other people thought Anna was not ready.
(Lots of people)
At 2 ½ years joint attention was less than 2
minutes.
20 minutes of 30 minute session involved
Anna crying.
- Mom thought this was an escape behavior.
- Mom seemed to be always feeding her. She
  was grazing.
- She had extreme episodes of hyperphagia.
  - Attempted extremity feedings were unsuccessful.

Her crying episodes decreased but never
went away.
- We learned this was from pain due to possible GI
  pain.
- IFSP included PT and SLP. Childhood
  Specialist but not Speech Therapy.
- Seizures/startting spells

ANNA

ANNA EXAMPLE
Expect changes over time; Don’t Quit

After the regression stage, individuals with RTT are reported to be sociable and to be responsive to their environment. Such characteristics may help sustain a conducive social environment.
THANK YOU!

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RESOURCES


https://www.rettsyndrome.org/
http://rettuniversity.org/
http://praacticalaac.org
https://thinksmartbox.com/product/look-to-learn/
http://complexneeds.org.uk/