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# Sotos Syndrome: Best Practices & Recommendations in Speech-Language Pathology



Philadelphia

I have no disclosures.

# Learner Outcomes

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Describe the SLP's role in diagnosing and managing Sotos syndrome



Identify at least 2 speech-language difficulties common in Sotos syndrome



Label at least 3 diagnostic measures for assessment



Label at least 3 key clinical management strategies



Label at least 3 key components of caregiver education and training

# What is Sotos Syndrome?

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## Overgrowth syndrome

- Intragenic variants in the NSD1 gene - at least 90% of cases
- 5q35 microdeletions of the NSD1 gene

# What is Sotos Syndrome?

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## Cardinal Features

Overgrowth (height and head circumference  $\geq 2$  SD above the mean)

Distinctive facial features

Neurodevelopmental features: intellectual disability (variable range), delayed development, behavioral challenges, hypotonia

# What is Sotos Syndrome?

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## Neurodevelopmental disorder

- Group of characteristics that may overlap with other disorders
- Onset during the developmental period and persist over a person's lifespan
- Range of developmental differences that vary from...
  - ...specific limitations to global impairments
- Wide diversity of functional impact

# Consider: Sotos and \_\_\_\_.

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## Sotos & Intellectual Disability

- Consider language development in relation to general intellectual development
- Useful tool – when available!

## Sotos & Autism Spectrum Disorder

- Clinically significant behavioral symptomology associated with ASD
- Social relations > restricted interests
- Forming and maintaining peer relationships

# Communication Challenges in Sotos Syndrome

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## Variability Across Individuals

Expressive language – semantics, syntax, grammar

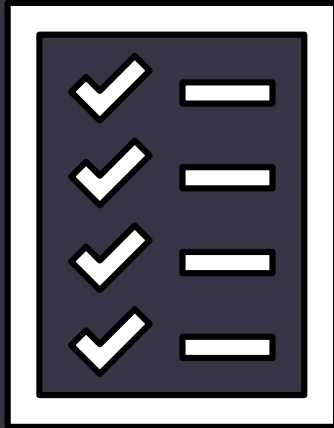
Social communication – social context, nonverbal communication

Receptive language – complex sentence structures & concepts

Literacy concerns

Articulation





# Diagnostic Tools

# Key Diagnostic Tools for SLPs

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## Diagnostic measures may include...

Caregiver Report

Criterion-Referenced Testing

Norm-Referenced Testing

Language Sampling

Dynamic Assessment

Which of these  
measures do you use  
most in your practice?

# Key Diagnostic Tool: Caregiver Report

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Caregiver report is essential – ask the critical questions!

- What are your concerns?
- Early language milestones?
- Other language exposures?

# Key Diagnostic Tool: Caregiver Report

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Caregiver report is essential – ask the critical questions!

- Family history of speech-language problems? Academic problems?
- Current services? Previous services?
- How can I, as the outpatient SLP, help?

# Key Diagnostic Tool: Caregiver Report

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Caregiver report – what if it's not an option?

- Review the medical record
- Ask the open-ended questions:
  - Can you give a few examples of your child's best communication?
  - How do you know that your child is smart? Bright? Clever?

# Key Diagnostic Tool: Criterion-Referenced Testing

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Pre-determined set of criteria

Range of developmental norms within each age range

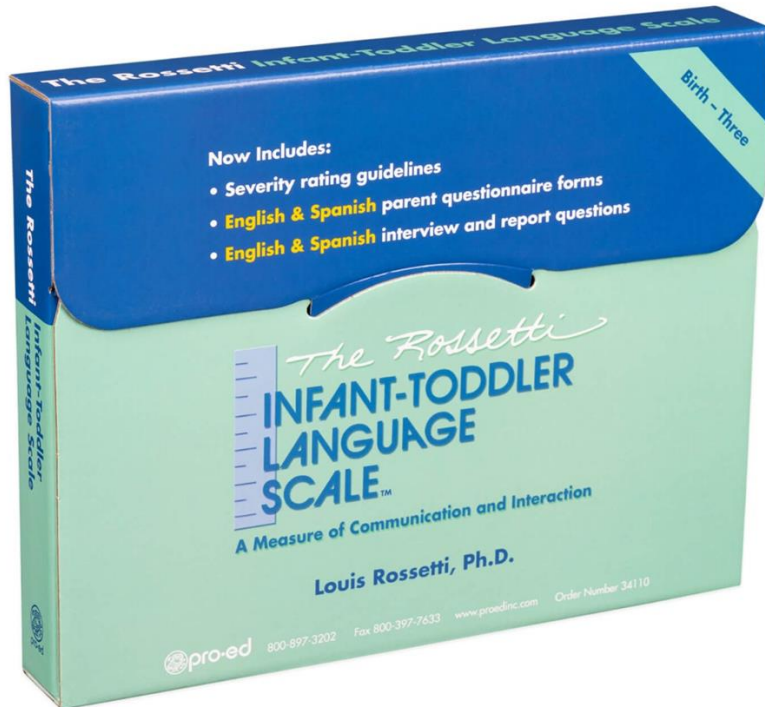
Allows quick insight into strengths & weaknesses

Easy to anticipate what goals come next

Good for tracking an individual's skill gains

Flexible administration

# Key Diagnostic Tool: Criterion-Referenced Testing



Current Communication Matrix Profile

MatrixID: 15305

Not Used

Emerging

Mastered

Surpassed

Level 1 Pre-Intentional Behavior	A1 Expresses Discomfort	A2 Expresses Comfort					A3 Expresses Interest in Other People										
Level 2 Intentional Behaviour	B1 Protests	B2 Continues Action		B3 Obtains More of Something			B4 Attracts Attention										
Level 3 Unconventional Communication	C1 Refuses, Rejects	C2 Requests More Action	C3 Requests New Action	C4 Requests More Object	C5 Makes Choices	C6 Requests New Object	C8 Requests Attention	C9 Shows Affection									
Level 4 Conventional Communication	C1 Refuses, Rejects	C2 Requests More Action	C3 Requests New Action	C4 Requests More Object	C5 Makes Choices	C6 Requests New Object	C8 Requests Attention	C9 Shows Affection	C10 Greets People	C11 Offers, Shares	C12 Direct Your Attention	C13 Polite Social Forms	C14 Answers Yes/No Questions	C15 Asks Questions			
Level 5 Concrete Symbols	C1 Refuses, Rejects	C2 Requests More Action	C3 Requests New Action	C4 Requests More Object	C5 Makes Choices	C6 Requests New Object	C7 Requests Absent Objects	C8 Requests Attention	C9 Shows Affection	C10 Greets People	C11 Offers, Shares	C12 Direct Your Attention	C13 Polite Social Forms	C14 Answers Yes/No Questions	C15 Asks Questions	C16 Names Things/ People	C17 Makes Comments
Level 6 Abstract Symbols	C1 Refuses, Rejects	C2 Requests More Action	C3 Requests New Action	C4 Requests More Object	C5 Makes Choices	C6 Requests New Object	C7 Requests Absent Objects	C8 Requests Attention	C9 Shows Affection	C10 Greets People	C11 Offers, Shares	C12 Direct Your Attention	C13 Polite Social Forms	C14 Answers Yes/No Questions	C15 Asks Questions	C16 Names Things/ People	C17 Makes Comments
Level 7 Language	C1 Refuses, Rejects	C2 Requests More Action	C3 Requests New Action	C4 Requests More Object	C5 Makes Choices	C6 Requests New Object	C7 Requests Absent Objects	C8 Requests Attention	C9 Shows Affection	C10 Greets People	C11 Offers, Shares	C12 Direct Your Attention	C13 Polite Social Forms	C14 Answers Yes/No Questions	C15 Asks Questions	C16 Names Things/ People	C17 Makes Comments
	Refuse	Obtain					Social					Information					

# Key Diagnostic Tool: Norm-Referenced Testing

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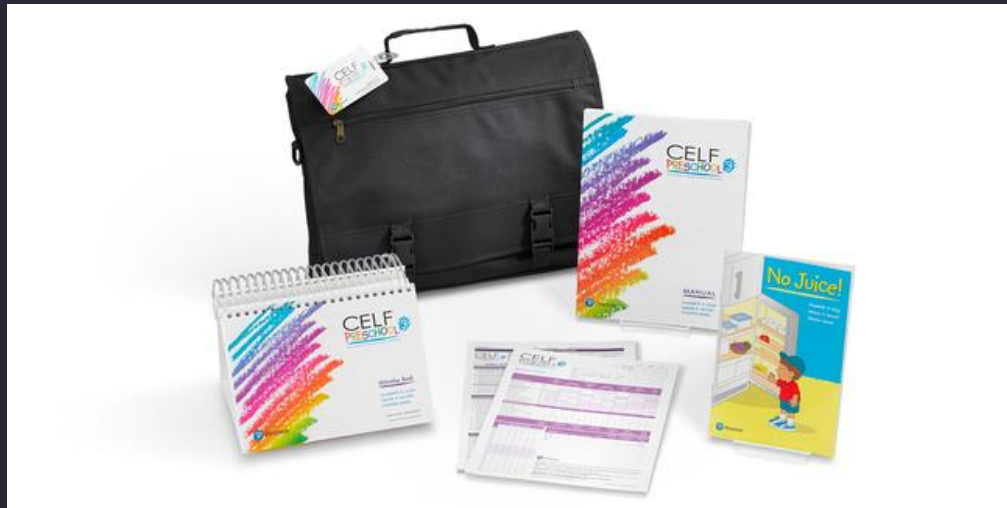
Compare and rank individuals with those of a large group of statistically selected individuals

Standard scores are derived, and performance is reflected on a bell curve

Provides an estimate of how far above or below the mean the individual performed



# Key Diagnostic Tool: Norm-Referenced Testing



# Key Diagnostic Tool: Language Sampling

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Assessing communication in a natural context

Grammatical development; Mean length of utterance; Use of language for daily functions

Alternative for: Multilingual or multidialectal patients; Individuals with behavioral differences that may limit participation

Contexts can include: Free play; Conversation; Narration; Expository speech

# Key Distinctions in Language Sampling

Language sampling can take the form of self-designed protocols & standardized protocols.

Standardized protocols include:

- Brown's Stages of Morphological and Syntactical Development
- SALT

If using a standardized protocol, language sampling may be considered a criterion-referenced assessment tool.

# Key Diagnostic Tool: Dynamic Assessment

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Used to identify present skills & learning potential (or, modifiability)

Mediated learning experience that involves scaffolding, explicit instruction and exposure to an identified area of need

Teach-test-retest model allows for differential diagnosis – language difference versus disorder!

Supports culturally and linguistically diverse learners

# Diagnostic Challenges

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**Norm-referenced tests** –  
may not be useful when a  
student is not represented in  
the normative sample



**Criterion-referenced tests** –  
contain bias for developmental  
norms; many agencies want  
standard scores



**Language sampling** –  
time consuming; requires  
training; requires threshold of  
utterances



**Dynamic Assessment** – may  
not be billable; often confused  
with Response to Intervention



# Treatment Planning

# Treatment Planning

**Per ASHA, treatment plan should encompass:**

Evidence-Based Practice

Individualized approach & Contextual factors

Interprofessional collaboration

Caregiver Education & Empowerment

# Treatment Planning: EBP

Treatments should be informed by latest research and evidence

- Clinical expertise: knowledge and critical reasoning acquired via clinical experiences
- Evidence: available information from scientific literature and from clinical observations
- Patient/caregiver perspectives: personal and cultural circumstances, values and priorities





# Treatment Planning: Individualized & Contextual

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Goals should be based on a comprehensive assessment

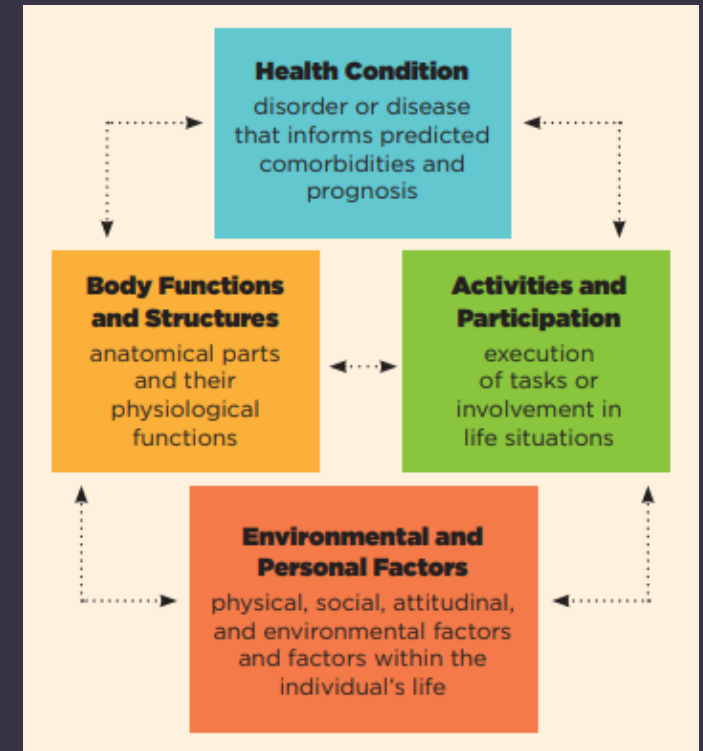
- Tailored to individual's needs and abilities
- Acknowledge and address cultural and linguistic differences
- Consider home, school and community environments
- Recognize the impact of social factors, such as SES, access to healthcare, etc.

When we think of person-centered treatment, we should be thinking about...

# Treatment Planning: Individualized & Contextual

## International Classification of Functioning, Disability and Health (ICF)

- Gives a voice to child and family
- Optimizes potential for child to participate in meaningful activities – which optimizes outcomes and leads to functional improvements



# Treatment Planning: Collaboration

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## Team approach – essential!

- Clinicians, therapists and educators are actively involved in patients care
- Increases patient- and family-informed decision making
- Enhances patient and family outcomes and yields diagnostic precision

# Treatment Planning: Caregiver Education & Empowerment

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Caregiver Education & Empowerment – essential!

- Enhances generalization of skills
- Ensures functional use of strategies at home

How do you engage caregivers in practice?

# Treatment Planning: Caregiver Education & Empowerment

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## Three Key Components

Triadic Model

Transactional Approach

Caregiver Competence & Confidence

# Caregiver Ed & Empowerment: Triadic Model

## Relational Practice

Compassion, active & reflective listening.

"Have you had a chance to review your child's initial speech-language evaluation?"

"Do you feel your child was their best self during that evaluation?"

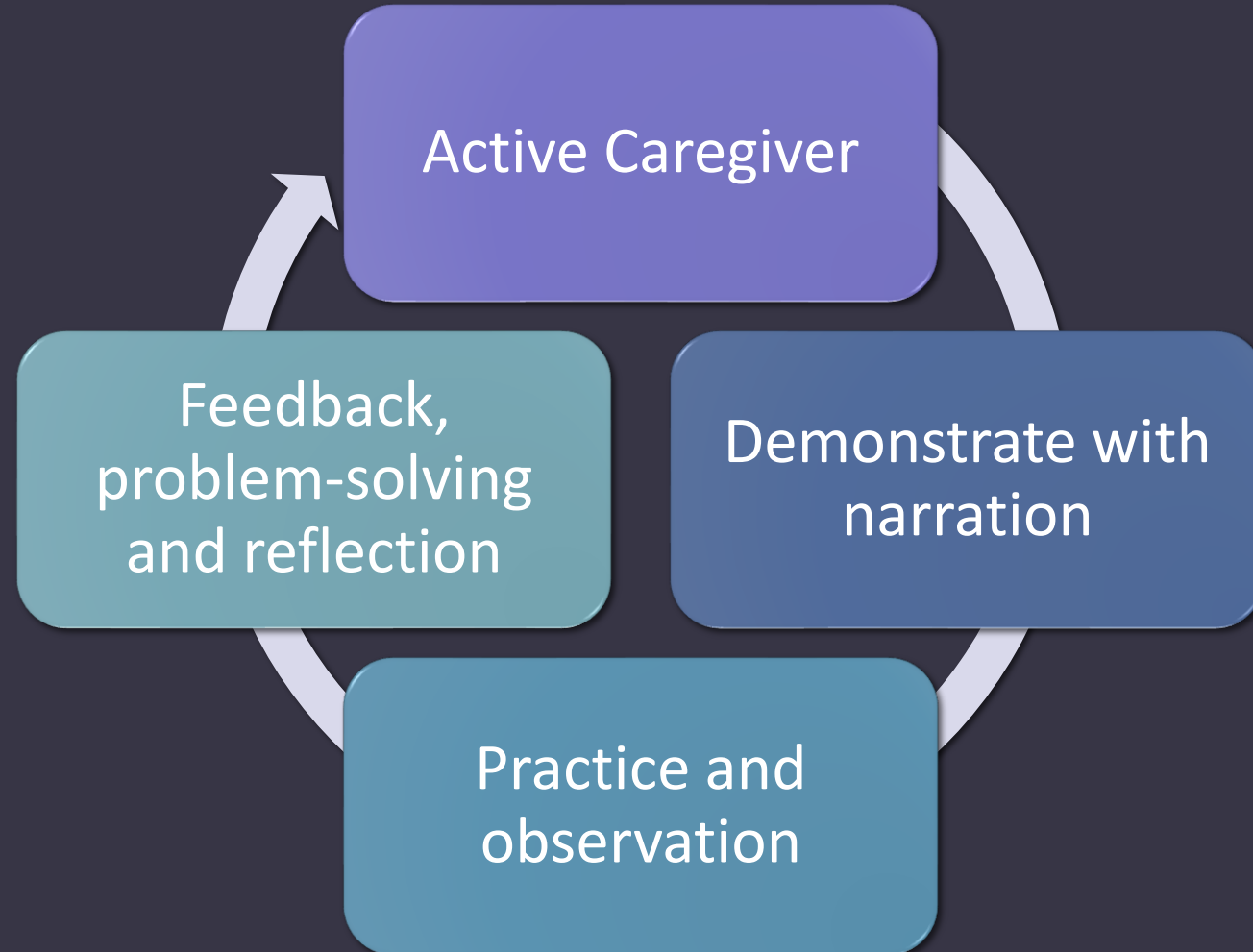
## Participatory Practice

Involve caregivers as decision- and change-makers.

"What do you think about these goals?"

"Can I show you how these goals relate to your child's current skills?"

# Caregiver Ed & Empowerment: Transactional Approach



# Caregiver Ed & Empowerment: Confidence & Competence

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Never assume competence!

- Reinforce caregivers' knowledge of treatment plan and intervention strategies.

Bolster caregiver's confidence to implement strategies in daily routine.

- SLP coaches the caregiver using a transactional approach.
- Equips the caregiver with appropriate techniques to continue those learning opportunities at home, with confidence.



# So, what do we know?

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SLP's play an integral role in the diagnosis and management of speech-language disorders in Sotos syndrome



It is important to be mindful of the intersectionality of Sotos syndrome



SLP's can utilize a variety of diagnostic measures to obtain a comprehensive estimate of a child's speech and language skills



Treatment plans should be evidence-based, individualized and contextual, and emphasize interprofessional collaboration



Caregivers play an essential role in their child's treatment plan – education and training is critical!

# Case Study



# Case Study: Introduction

## Initial Presentation

AB is a 2 year, 8-month-old  
Followed by Overgrowth Clinic at CHOP for a  
diagnosis of Sotos syndrome (5q35  
deletion).

## Chart Review

Additional diagnoses: autism spectrum  
disorder; global developmental delay and  
right hemiparetic cerebral palsy;  
oropharyngeal dysphagia, aspiration and  
GERD.

## Caregiver Interview

AB receives speech-language therapy 1x/week through early intervention and 1x/week  
through private center.  
Additional therapies: feeding therapy, ABA, occupational and physical therapies.  
Family goals: looking for a functional and effective communication system to reduce  
communication breakdowns; vocabulary expansion.

Which diagnostic tools would you  
prioritize?

# Case Study: Evaluation

## Testing Measure(s)

Criterion-referenced assessment tool: Communication Matrix for Professionals

Play & Observation

**The Communication Matrix for Professionals (2004); Charity Rowland, Ph.D.**

Reason to Communicate	Primary Level of Communication Competence	Highest Level of Communication Competence
To Refuse	Level 3: Unconventional Communication	Level 4: Conventional Communication
To Obtain	Level 3: Unconventional Communication	Level 6: Abstract Symbols
To Socially Engage	Level 3: Unconventional Communication	Level 4: Conventional Communication
To Provide or Seek Information	Not yet applicable	Level 4: Conventional Communication

# Case Study: Evaluation Results

## Receptive Language Profile

- Comprehension of routine and familiar directions
- Identification of familiar people and objects
- Does not yet follow novel directions consistently
- Working to expand vocabulary knowledge

## Expressive Language Profile

- Strong communicative intent
- Limited functional communication
- Relies primarily on:
  - Physical manipulation
  - Small repertoire of gestures and signs
- Emergent use of canonical and variegated babbling

# Case Study: Evaluation Results

## Social Communication

- Demonstrates reciprocal interaction
- Shows shared enjoyment and affection
- Excellent imitation skills during play
- Working on expanding participation in varied play routines

## Speech Sound Development

- Presents with limited speech sound inventory
- To be monitored for future goal development

What would be your top  
treatment priorities?



# Case Study: Conclusions

## Recommendations

Continue speech-language therapy through the EI and privately.

Focus on:

- Functional vocabulary development
- Following directions
- Expansion of imitation skills and use of supplemental gestures
- Participation in a variety of play routines

# Case Study: Conclusions

## Recommendations

### AAC Evaluation

- An occupational therapist trained in assistive technology (AT) should be present to support assessment of access methods and device use.
- A co-evaluation (AAC/AT) at CHOP is recommended to determine the most appropriate communication equipment for AB.
- Caregivers completed an intake form and will be contacted for scheduling.

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# Thank You!

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