THE DUAL DIAGNOSIS OF DOWN SYNDROME AND AUTISM

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Webinar for The Matthew Foundation

DISCLOSURE

No one involved in the planning or presentation of this activity has any relevant financial relationships with a commercial interest to disclose.

WHAT IS DOWN SYNDROME?

- A genetic syndrome
- Caused by an extra copy of the 21st chromosome
- Typically diagnosed at birth based on facial and body features
- Diagnosed with a blood test

WHAT IS AUTISM?

- A developmental difference
- Causes of autism are unknown at this time
- 3 main features:
  - Difficulties with communication, especially social communication
    - Also includes difficulties understanding and sustaining social interactions
  - Restrictive and repetitive patterns of interests and behaviors
  - Sensory integration difficulties
- Diagnosed around 2-4 years old, sometimes later
- Diagnosed with a full developmental evaluation
  - There is no blood test that can diagnose autism

CAN DOWN SYNDROME AND AUTISM OCCUR TOGETHER?

- Until recently, it was believed that ASD is rare in individuals with Down syndrome
- Because children with Down syndrome have been described as friendly and affectionate, this relative strength in social skills was thought to be protective against autism.
- We now know that a lot of people with Down syndrome also have autism
- It is important to recognize when someone has both Down syndrome and autism, so that we can best support their learning

OUR PRESENTATION TODAY

Our goal today is to share information about children with a dual diagnosis of Down syndrome and Autism.

This information was gathered in two ways:

- A review of existing literature on the topic
- Original research designed by the DSMIG DS-ASD workgroup and completed by our Down syndrome clinic

RESEARCH SURVEY

- Developed to understand the experiences of families with children with DS-ASD
  - Early years prior to diagnosis,
  - Interactions with the medical and education systems in obtaining a diagnosis
  - Perspectives on what interventions and supports have been helpful
  - Reflections on how the dual diagnosis has affected the family unit.

EPIDEMIOLOGY

- ~16% of people with Down syndrome also have autism
- It is more common in males
- It may be more likely with some medical co-morbidities, especially infantile spasm
- It is associated with lower IQ scores
- Sometimes associated with a family history of developmental issues

PRESENTATION

FROM SYMPTOMS TO DIAGNOSIS

Age in Years				
Category	**Mean**	**Median**	**Range**	**SD**
Age at First Concern	3.87	3.00	0.25 - 12.00	2.56
Age at Diagnosis	8.52	7.00	1.50 - 21.00	4.77
Years Between First Concern and Diagnosis ("Age Gap")	4.65	3.50	0.00 - 17.50	3.67

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PRESENTATION – POINT OF FIRST CONTACT

Who Caregivers Reported Concerns to, and Whether they had the Knowledge to Guide Further Evaluation			
Recipient of Concern	**Frequency**	**Knowledgeable**	**Not Knowledgeable**
Family	6 (13%)	0 (0%)	6 (100%)
Educator / Therapeutic Provider	27 (57%)	6 (22%)	21 (78%)
Pediatrician / PCP	22 (47%)	4 (18%)	18 (82%)
Medical Specialist	12 (26%)	10 (83%)	3 (25%)

HOW CHILDREN PRESENT

Survey question:

What were some things you noticed about your child that made you think they were a little different from other children with Down syndrome?

STEREOTYPIC BEHAVIORS

- Repetitive behaviors (e.g. hand flapping, stimming, head banging)
- Perseverative behavior (e.g. staring at ceiling fans or lights, playing with same toy over and over)
- Children with DS have elevated rates of repetitive/stereotypical movements
  - 60% of kids with DS without Autism have elevated "mannerisms" scores
- A subset of patients with DS who have stereotyped movements/behaviors have ASD
- **Children with DS-ASD do it more, it's more complex, and it's harder to redirect them**

ABNORMAL PLAY

- Lining up toys
- Sorting instead of playing with the toy
- Limited pretend play
- Playing with a toy in the same way over and over
- Focusing on part of a toy instead of the whole toy
- Older children: continuing to put toys in the mouth

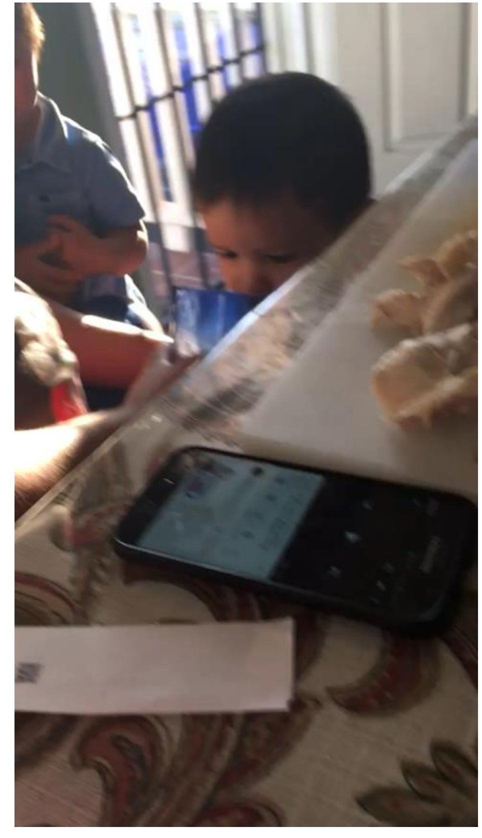
IMPAIRED SOCIAL SKILLS

- Still more social than kids with autism without Down syndrome
- The interest in social interaction is less sustained
- Less imitation, back and forth
- Less calling attention to what the kid is doing, “shared attention”
- Not joining others in play, prefers to play alone
- Less interested in socializing, more withdrawn

MARKED COMMUNICATION DIFFICULTIES

- Limited verbal and non-verbal communication
- Acquire language later
- Difficulties with both receptive language (understanding what others are saying) as well as expressive language (speaking)
- Repetitive speech/echolalia (repeating the last thing someone said)
- May still repeat parts of a song, movie, but out of context

EXPRESSIVE LANGUAGE DELAY, NOT AUTISM



SENSORY DIFFICULTIES

- Significant feeding difficulties
- Prefer to be isolated in low sensory environments
- Refuse touch-type things especially creams, lotions, drops, hair cuts
- Difficulty with loud noises, darkness, crowds
- Does not like being touched by others
- Crowds, laughing too loud, too many people talking
- May have tantrums that are unexplained and are difficult to soothe

POOR EYE CONTACT

- Most of my patients still look at me in the eyes
- However, the eye contact is not sustained, and usually not communicative
  - More of a “checking in that you are here”
- May focus on my mouth instead
- May give me a side gaze

SLOWER ACHIEVEMENT OF MILESTONES

- Progressing more slowly than other children with Down syndrome
- Communication most affected
- Fine motor, self help skills also affected
  - Greater delays in toilet training, self feeding

MOOD AND BEHAVIOR ISSUES

- Tantrums
- Hyperactivity
- Impulsivity
- Self-injury
- Aggression
- Anxiety
- Not soothed by physical contact or verbal comforting

DEVELOPMENTAL REGRESSION IN EARLY CHILDHOOD

- More children with DS+ASD have a history of regression in language and general skills than children with DS-Only (Warner et al., 2014; Castillo et al., 2008).
- About 1/3 of children with DS+ASD have a history of regression (Capone et al, 2006).
  - Rate comparable with that reported in idiopathic ASD
- Occurs later (Castillo et al., 2008).
  - Between ages 4-5 years for DS+ASD vs <2 years for idiopathic ASD
- Make sure you rule out medical causes of regression – seizures, hypothyroidism, systemic disease

TAKE HOME

- Concern may be "my child is different from other kids with Down Syndrome"
- Signs tend to be noticed around 3-5 years old
- Families may have a hard time getting their concerns heard
- Symptoms reported are symptoms of Autism
  - Stimming more complex and hard to redirect
  - May look social but act less social (ex prefers to play alone)
  - Significant language delays: receptive, expressive, gestural
  - May have hyperactivity, anxiety

EVALUATION

PAUCITY OF EVIDENCE

There is "insufficient research-based guidance on the selection or use of reliable and valid ASD screening measures for children with DS"
(Bull & The Committee On Genetics, 2011).

THE BASICS

- **Hearing screen** at 6 months, 12 months, and every 6 months thereafter until ear-specific information can be obtained
- **Eye check:** Check red reflex at every check up 0-12 months, congenital cataracts can develop slowly in DS. **Ophthalmologic** evaluation by 12 months of age, then yearly optometry/ophthalmology until school age
- **Thyroid** at 6 months, 12 months, yearly thereafter

MCHAT-R/F

- Modified Checklist for Autism in Toddlers-Revised, with Follow-Up (Robins, Fein & Barton, 2009) – **every pediatrician's office has this**
- Parent-report screener for toddlers 16-30 months of age
  - Step 1: 20-item screener
  - Step 2: follow-up questions for failed items
- In verbal children with DS under 60 months of age, and in nonverbal children with DS under 72 months of age, DiGiuseppi et al. 2010 found a sensitivity of 81.8% and specificity of 46.8%

AUTISM DIAGNOSTIC OBSERVATION SCHEDULE - ADOS

RECOMMENDATIONS FOR ASSESSMENT

- Parent interview based on ADI-R
- Teacher information about social interactions with peers
- Social Communication Questionnaire (SCQ)
- Aberrant Behavior Checklist (ABC)
- Vineland
- Autism Diagnostic Observation Schedule (ADOS-2)

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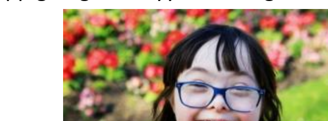
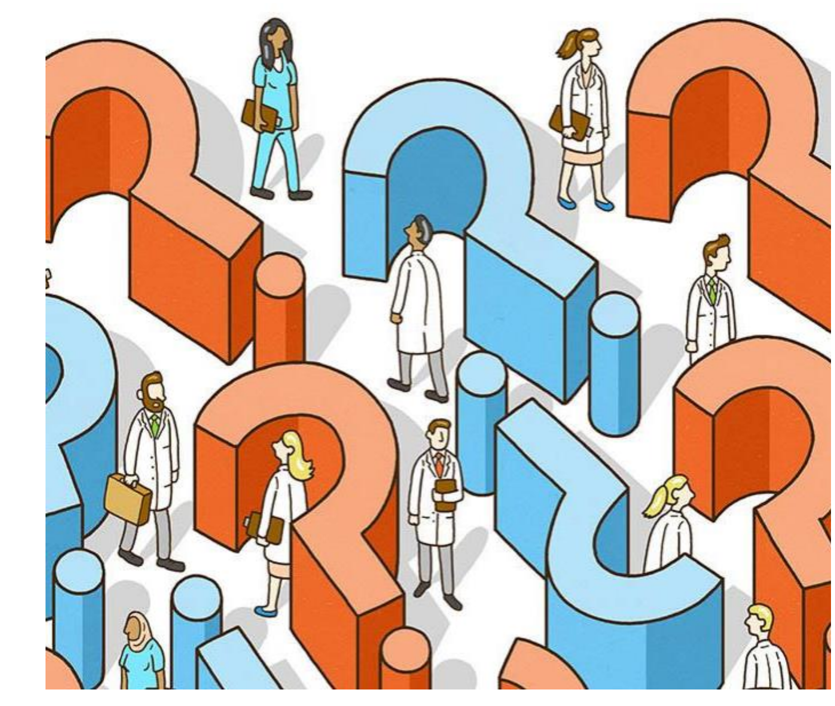


INTERVENTION

PAUCITY OF EVIDENCE

- Almost no scientific research on treatments specific for DS-ASD and associated problems.
- In children with DS-ASD, autism is the primary disability that interferes most with learning and behavior.
- Intervention is, therefore, strongly based on work with children who have autism.



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CHALLENGES FOR CHILDREN WITH DS-ASD

Perceived Weaknesses of Child with DS-ASD	
Communication Deficits	57%
Rigidity	41%
Impaired Social Skills	30%
Mood and Behavior Issues	25%
Dependent on Caregiver for ADLS	23%
Sensory Issues	16%

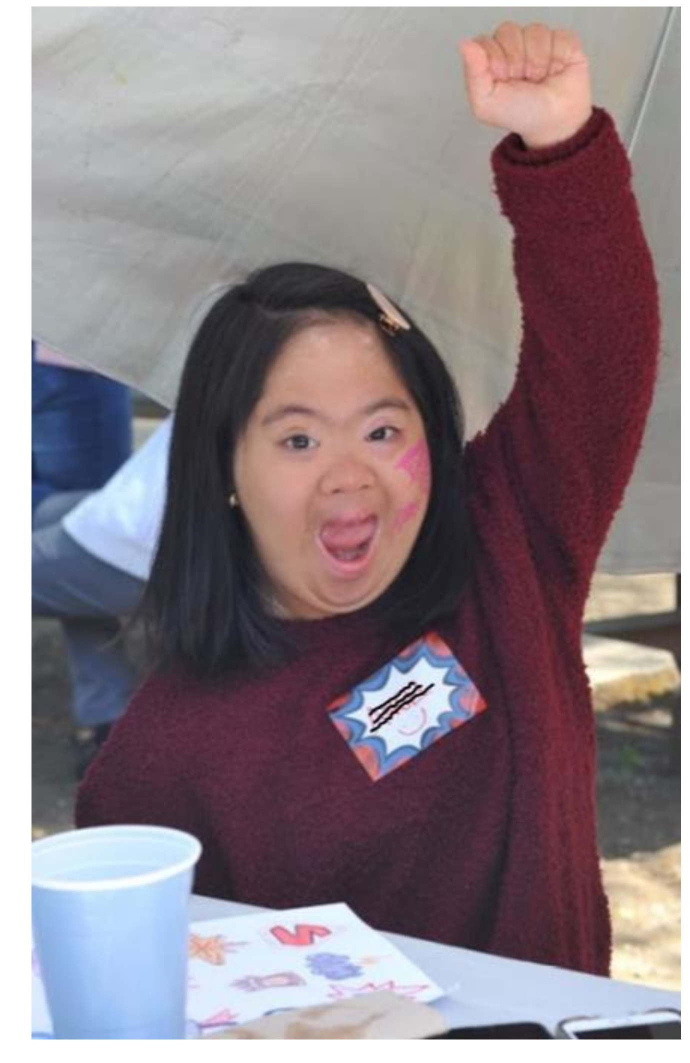
Perceived Weaknesses of Child with DS-ASD	
Safety Concerns	16%
Attention Issues	14%
Cognitive Skills	11%
Gross and Fine Motor Delays	7%

GOAL SETTING

1. Maintain a long-term perspective
   - Quality of life, Independence, Safety

1. Identify goals that support the long-term outcome
   - Communication, Social functioning, Self-care

1. Shorter term objectives
   - Attending, compliance, waiting, compliance, social skills



FACTORS RELATED TO OUTCOME

Characteristics of the intervention

- Dosing
  - Intensity
  - Duration
- Application of intervention
  - Consistency
  - Targeting the right thing
- Situational factors
  - Concurrent stresses
  - Lack of resources

Characteristics of the child

- Cognitive abilities
- Language abilities
- Adaptive functioning
- Imitation, joint attention
- Other co-morbidities

HELPFUL INTERVENTIONS

Helpful Interventions for Child
Applied Behavioral Analysis
Physical Therapy, Occupational Therapy, and Speech Therapy
School
Parental Support
Down Syndrome or Autism Clinics
Recreational Activities
One on One Support
Communication Interventions
Psychological and Medical Services

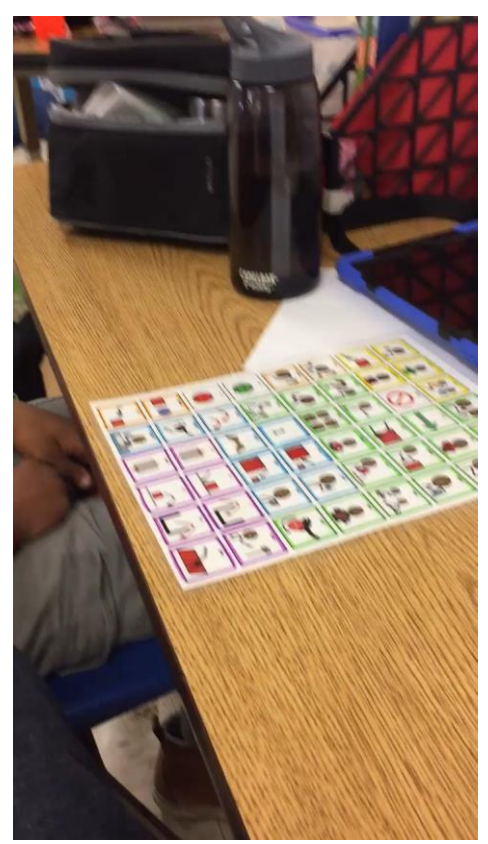
ABA

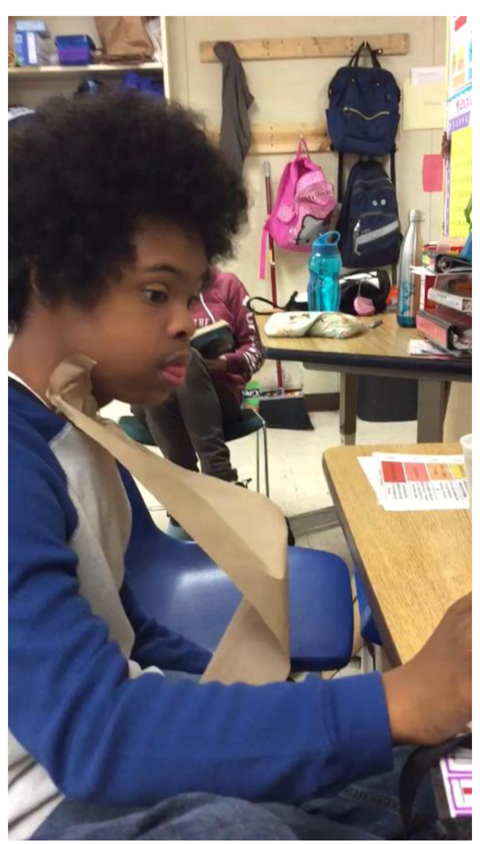
https://www.youtube.com/watch?v=OKzYzsDQCiY

PARENT TRAINING

- Based on principles of Applied Behavior Analysis (ABA)
- Treatment includes 11 Core sessions, 7 Supplemental sessions, a home visit, and follow-up telephone booster sessions.
- Sessions contain a therapist's script, activity sheets, a parent handout, and treatment fidelity checklists. Video vignettes demonstrate concepts taught in session
- Feasible and effective. The majority of the children demonstrated improvements in problem behavior. (Edwards, Zlomke, Greathoue, 2019).
- In large scale RCTs, structured parent training was superior to parent education for reducing disruptive behavior in children with ASD (Bearss et al., 2015).
- PT confers additional benefit when combined with medication (Aman et al., 2009; Scahill et al., 2012)

SUPPORTING COMMUNICATION

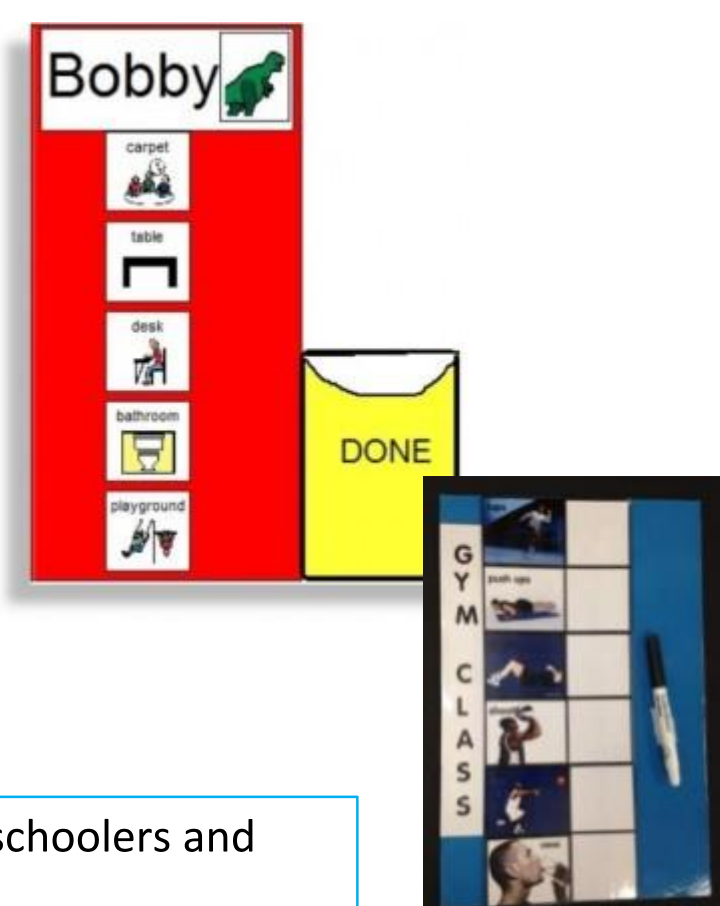




STRUCTURED TEACHING - TEACCH

Five principles:

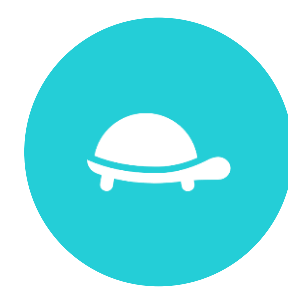
- Physical structure – Layout of surroundings
- Scheduling – What to do, when to do it. Presented in multiple ways.
- Work System – What is expected of the individual/what is to be accomplished; what happens after activity is completed
- Routine – Checking schedule and following system
- Visual structure – visual based cues
- Siua AMH, Linb Z, Chung J. 2019 (adults with ASD/ID, + control group): TEACCH had significantly larger improvement in the goal attainment scaling scores than the comparison group



Other programs in ASD literature: Learning Experiences and Alternative Program for Preschoolers and Their Parents (LEAP), UCLA/Lovaas-based behavioral intervention

MEDICATIONS

Set clear goals



Start low, go slow

See the doctor often to monitor for effect and side effects

Whenever possible, consult with a child psychiatrist or developmental and behavioral pediatrician

MEDICATIONS

- No medication will “cure” autism
- Medication can help decrease behavioral symptoms such as hyperactivity, aggression, anxiety, self injury

Medication	Targets	Side Effects
Aripiprazole (Abilify)	Emotional distress, aggression, hyperactivity, self-injury	Marked weight gain, Drowsiness, Extrapyramidal symptoms (muscle stiffness or tremor)
Risperidone (Risperdal)	Aggression, hyperactivity, irritability	Weight gain, Sedation (drowsiness, especially at first), hyperprolactinemia
Methylphenidiate or Amphetamines	Hyperactivity, inattention, impulse control	Appetite suppression, Sleep disruption, Can be activating and cause irritability in some children
Clonidine, Guanfacine	Hyperactivity, inattention, self injury, sleep	Low blood pressure, sedation
SSRIs (fluoxetine, sertraline)	Anxiety	Akathisia, GI upset

TAKE HOME

- ABA may be helpful
- Parent training may be helpful
- School needs to be structured
- Offer communication support
- Refer to a Down syndrome clinic
- Consider medication

FAMILY

RECEIVING A DIAGNOSIS

- Frequently, parents of children with DS+ASD welcome their child’s additional diagnosis with relief, as they have often been grappling with why their own child does not conform to the stereotype of the friendly child with DS, and with why his/her progress has not kept pace with that of other children with DS
- The diagnosis of Autism can help to explain those differences, and can offer hope that a shift in approach to the child’s special services will confer more significant gains in their development (Piper & Howlin, 1992).
- For some parents, however, the diagnosis represents another set of problems. They may feel more isolated than previously and not feel that they are part of any community.

RECOMMENDATIONS

DS-ASD RECOMMENDATIONS FOR CAREGIVERS

If you question whether your child with Down syndrome has autism

- List and videotape behaviors that make you question ASD
- Share your observations with your child's primary care provider, and get a second opinion if s/he is not concerned
- Seek a formal evaluation by a psychologist and/or developmental pediatrician to rule out ASD
- Understand that an additional diagnosis of ASD (and also ADHD, Stereotypic Movement Disorder, and Obsessive Compulsive Disorder) can allow access to more school-and community-based services
- Learn about ASD in Down syndrome ("dual diagnosis," "DS-ASD")
  - Search www.ndss.org for "autism"
  - *More Than Down Syndrome: A Parent's View*, by Joan Medlen (1999). Disability Solutions, Volume 3: Issue 5&6

Common tools used to evaluate for autism

- Screening measures: MCHAT, Social Communication Questionnaire
- Common parts of the evaluation: parent interview, developmental/cognitive testing, adaptive measure (Vineland, ABAS), structured observation (ADOS-2)

If your child with Down syndrome is diagnosed with autism

- Learn about autism in Down syndrome (DS-ASD)
  - www.ds-asd-connection.org
  - *When Down Syndrome and Autism Intersect: A Guide to DS-ASD for Parents and Professionals*, by Margaret Froehlke & Robin Zaborek (2013)
- Connect with community agencies
  - Local DS Guild/Associations
  - State Division of Developmental Disabilities
  - Seek out caregiver support groups, such as the Down Syndrome-Autism Connection Support private group Facebook page
- Learn about evidence-based treatment interventions and build your team
  - www.autismspeaks.org , www.parentcenterhub.org/find-your-center/
  - Identify speech, occupational, and physical therapists, behavior therapists (Behavior Analyst Certification Board website), educators and health professionals with experience with DS and ASD, and people who offer you emotional support
- Seek services through your school district
  - Seek early intervention (0-3 yrs) or early childhood special education (3-6 yrs)
  - Have autism listed as the primary disability on your child's IEP
  - Request an augmentative communication evaluation, if applicable
- Keep records
  - Challenging behaviors, adaptive skills, what works/what doesn't work

DS-ASD DSMIG Workgroup
Down Syndrome-Autism Connection
October 2019

Down Syndrome Connection of the Bay Area

UCSF Benioff Children's Hospital
Oakland

DOWN SYNDROME-AUTISM
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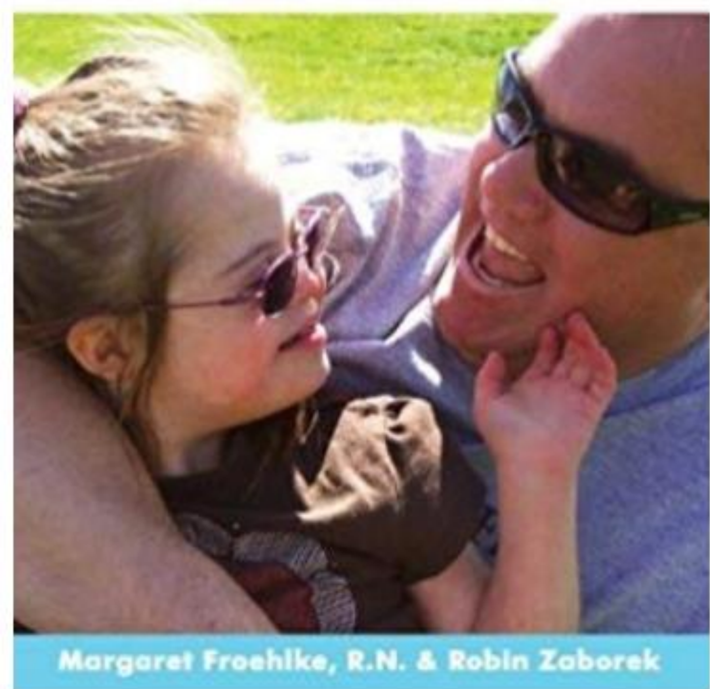
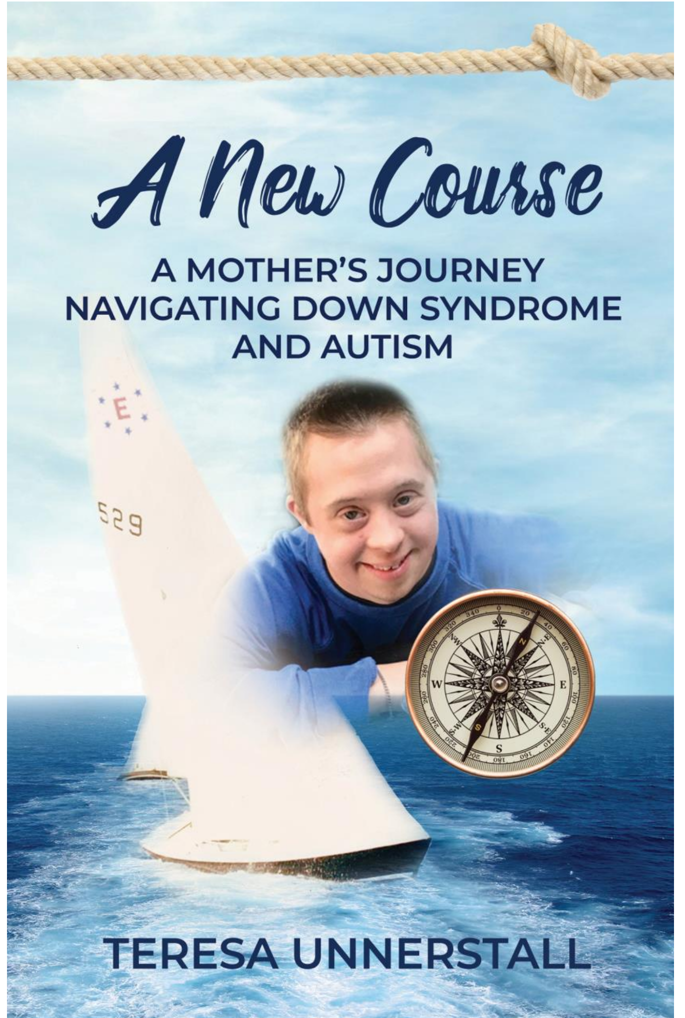
When **Down Syndrome** and **Autism Intersect**

A Guide to DS-ASD for Parents and Professionals



Margaret Froehlke, R.N. & Robin Zaborek



Down Syndrome & Autism

Down syndrome and autism are both equal opportunity conditions– meaning that anyone, anywhere, regardless of race, creed or socio-economic status, can have a child with either condition. It is believed that up to 18%[1] (some research suggests up to 39%[2]) of individuals with Down syndrome also have autism spectrum disorder. Both Down syndrome and autism can be challenging disabilities separately, without the combination, however when combined the challenges are multiplied and can be quite complex.

Behaviors to Notice

Before 3 years old:

- Repetitive motor behaviors
- Fascination with and staring at lights, ceiling fans, or fingers
- Episodic eye movements
- Extreme food refusal
- Unusual play with toys or other objects
- Receptive language impairment
- Little or no meaningful spoken language, gestures or signs

3 years and older:

- History of developmental regression
- Hyper or hypo-activity, short attention, impulsivity and poor organization
- Unusual vocalizations
- Unusual sensory responsiveness
- Difficulty with changes in routine or familiar surroundings
- Extreme anxiety, fearfulness or agitation
- Sleep disturbances
- Disruptive behaviors

Teens and Adults:

- Significant lack of social response or relatedness with family or friends
- Lack of interest or ability to develop relationships with peers
- Antisocial, anxious, or fearful in the presence of people they don't know
- Intensified stereotypic and repetitive motor behaviors
- Obsession or fascination with inanimate objects
- Lack of ability or interest in creative play
- Manipulation of objects in rigid ways
- Intensified sensitivity to certain types of sensory input
- Frequent tantrums and outbursts, as well as verbal or physical aggression
- Great difficulty in adjusting to transitions
- Dropping to the ground and refusing to move

Please note that many of these behaviors are normal for children with Down syndrome at certain points of development. Also, a child with Down syndrome may experience relatively normal development but then regress by developing these behaviors between the ages of three and seven. When one or two of these behaviors become predictable, extreme, or resistant to change, your child may benefit from a thorough evaluation for autism spectrum disorder performed by a professional who is experienced in working with children with Down syndrome.

1 JFK Partners, University of Colorado Denver, http://jfkpartners.org.

2 When Down Syndrome and Autism Intersect, A Guide to DS-ASD for Parents and Professionals, P.1, Woodbine House, 2013.

ACKNOWLEDGEMENTS

DSMIG-USA®
Down Syndrome Medical Interest Group-USA

- DSMIG-USA is a group of health professionals committed to promoting the optimal health care and wellness of individuals with DS across the lifespan.
- Members of this DSMIG-USA are professionals from a variety of disciplines who provide care to individuals with Down syndrome and/or their families. This may include physicians, scientists, psychologists, nurses, genetic counselors, educators, therapists, clinic coordinators, and related health professionals.
- Most members work in specialized Down syndrome clinics, at academic institutions, university- and community-based medical centers, or private practice.

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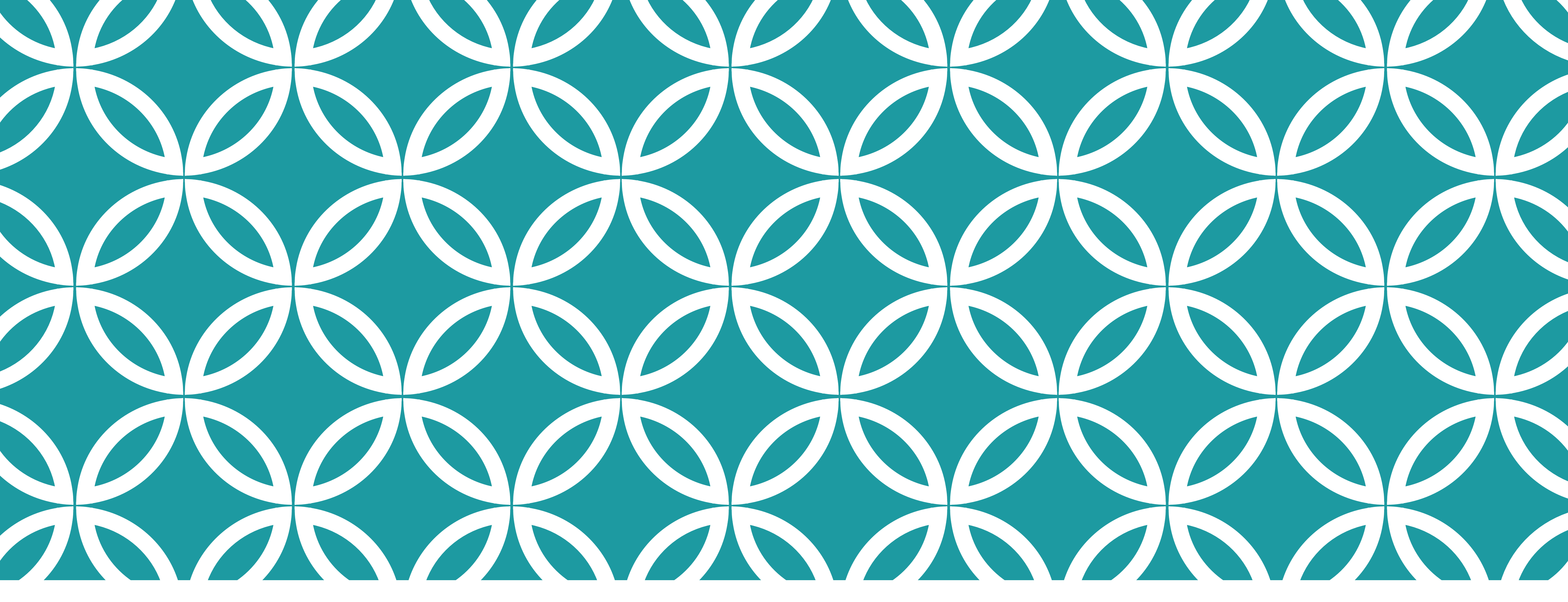

The Dual Diagnosis of Down Syndrome and Autism

A Parent's Perspective

Teresa Unnerstall, DS-ASD Consultant

Author of *A New Course: A Mother's Journey Navigating Down Syndrome and Autism*

www.teresaunnerstall.com

Nick Unnerstall, 27 years old with a dual diagnosis of Down syndrome and autism (DS-ASD)

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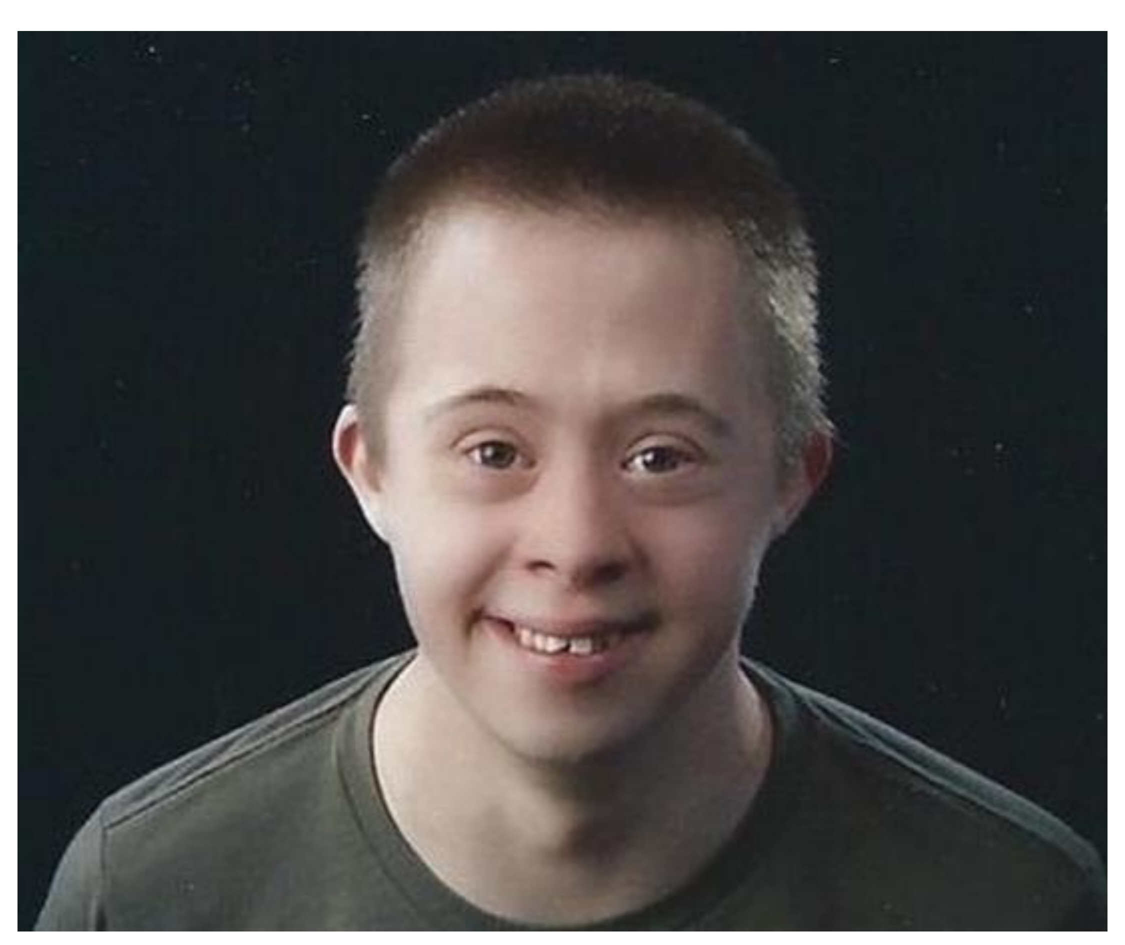

A Memoir with Chapter Lessons and a Full Appendix of Takeaways

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Overview and Objectives

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- Red flags and evaluation at age 5
- Red flags and evaluation age 10
- Working with the IEP team to address autism
- Helpful interventions: 4 key areas
- Finding supports and educational advocacy
- Utilizing strengths and minimizing barriers
- Putting it all together: A Transdisciplinary Approach

Red Flags at Age 5

- Stimming and sensory seeking
- Vocal stimming
- Disinterest in engaging and signing nursery rhymes
- Lack of speech development
- OT evaluation denotes characteristics of sensory integration disorder—sensory sensitivity with haircuts, textures like sand, clothing, etc…)
- Self-Injurious behaviors (SIBS)

Nick age 5 in Livermore, CA (2001)

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Nick's Evaluation for autism, age 5

- Evaluation done by a developmental pediatric psychiatrist ruled out autism.
- Parent interview/questionnaire and a brief observation of Nick interacting through play.
- No flip of the switch on language decreasing or regression of skills.
- Report indicated that Nick was highly social made good eye contact and the speech therapist evaluation of Verbal Apraxia of Speech was the cause of speech delays that should improve with continued treatment over time.

Red Flags age 10

- Increase in stimming and SIBS
- Frustration levels amped up
- Aggression and meltdowns at home and with school staff
- Puberty and hormonal changes
- Delays in toileting and speech
- Social interaction-GiGi's Playhouse, seems social, but no interaction or reciprocal play with peers
- Parents felt isolated, not fitting in to DS support groups anymore

We are in over our heads!

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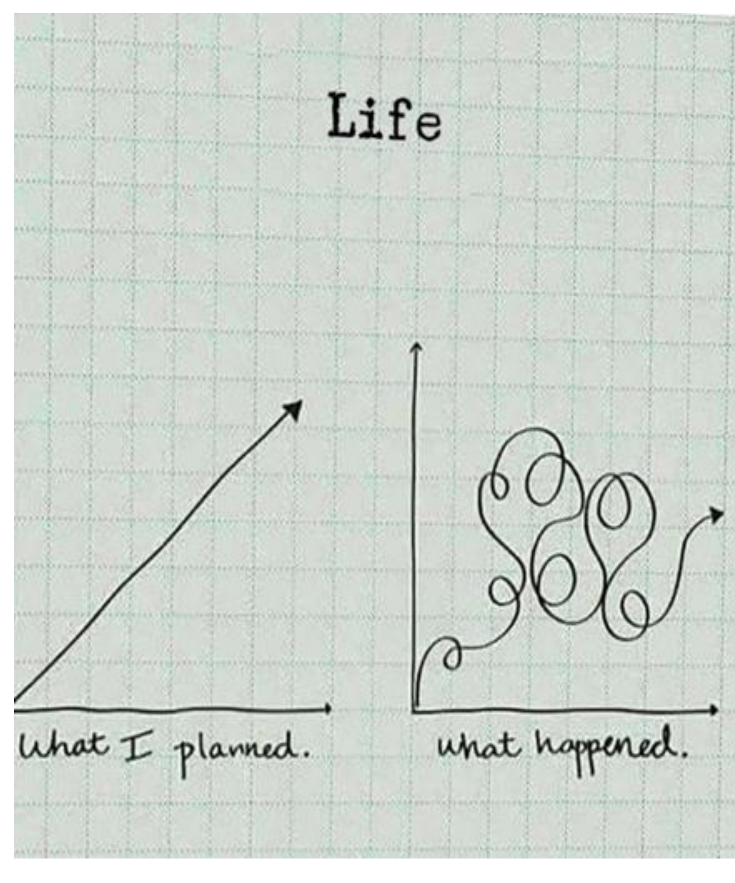

When I get frustrated

Now, what do we do?

- School District not willing to do an evaluation.
- Parents seek an independent evaluation
- Nick was given a secondary diagnosis of autism at age 11.

IEP Meeting called to address the secondary diagnosis of autism

- BCBA put on the team to do observations and make recommendations
- Functional Behavior Analysis (FBA) with data collection to create a positive behavior support plan (BSP)
- Staff/parent training in properly using a Picture Exchange Communication (PECS)
- Additional accommodations : Visual schedules, social stories, task strips, sensory breaks, transitions

“The secondary diagnosis of autism can unlock doors for more services and support to maximize a child’s potential.” *-Teresa Unnerstall*

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DS-ASD Areas Impacted

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DS-ASD

Areas Impacted

Behavior

Sensory

Helpful Interventions: 4 Key Areas

- **Speech and communication** evaluation for augmentative and alternative communication (AAC) and/or low tech and high tech picture exchange system (PECS).
- **Behavior/ Applied Behavior Analysis (ABA)** to address targeted behaviors.
- **OT collaboration to build in a sensory diet** to support the child.
- Apply for **state waiver funding** for in home family support for respite care, and home equipment to insure safety, diapers/adult diapers. **Supplemental Security Income (SSI)** for qualifying families.

Communication:
Visuals help to navigate smoother seas

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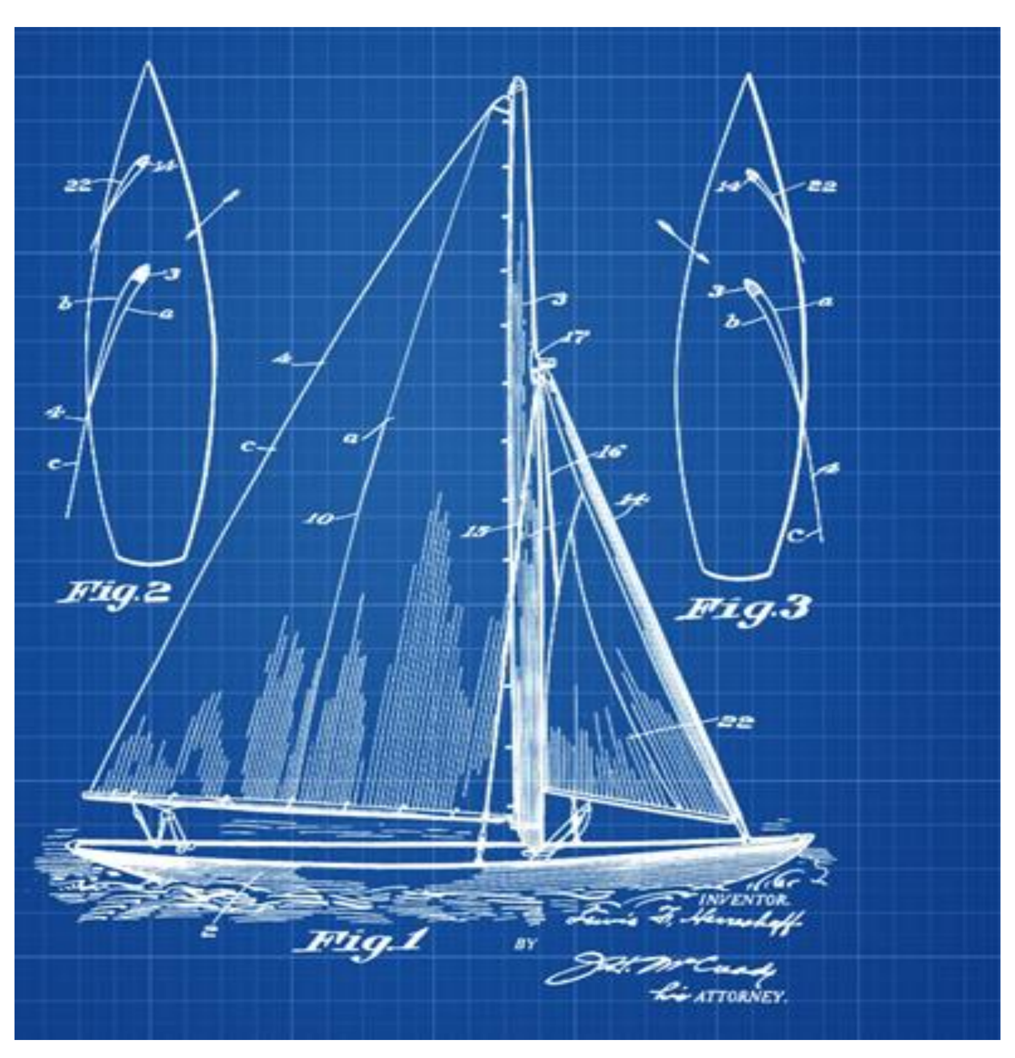

AAC: Low and High Tech using PECS

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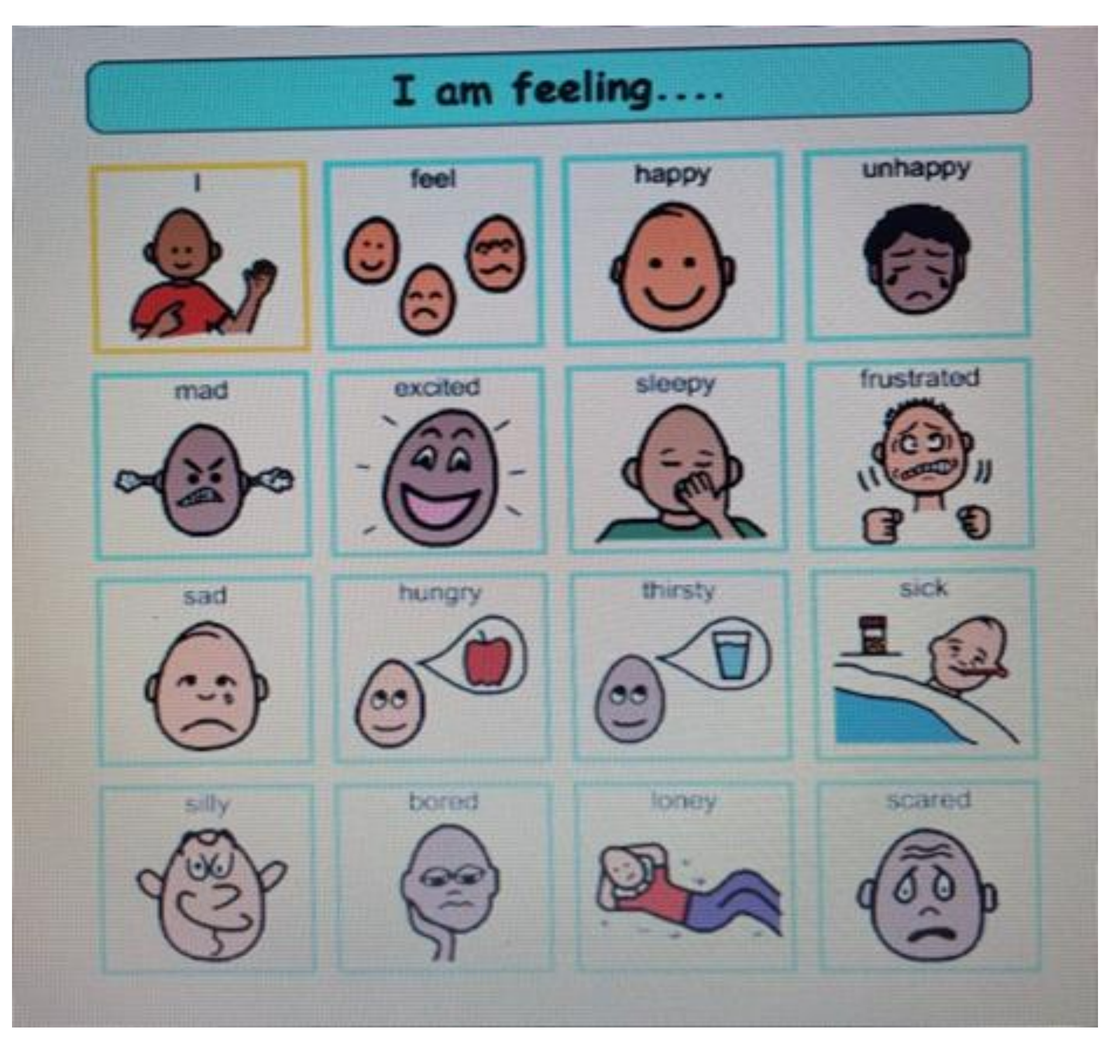
Low Tech Communication Library Visit: www.dscba.org



Visual Schedules provide predictability that will reduce anxiety!

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Task strips and social stories:

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Other Instructional Strategies:

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Nick using a visual schedule/ Timer Apps

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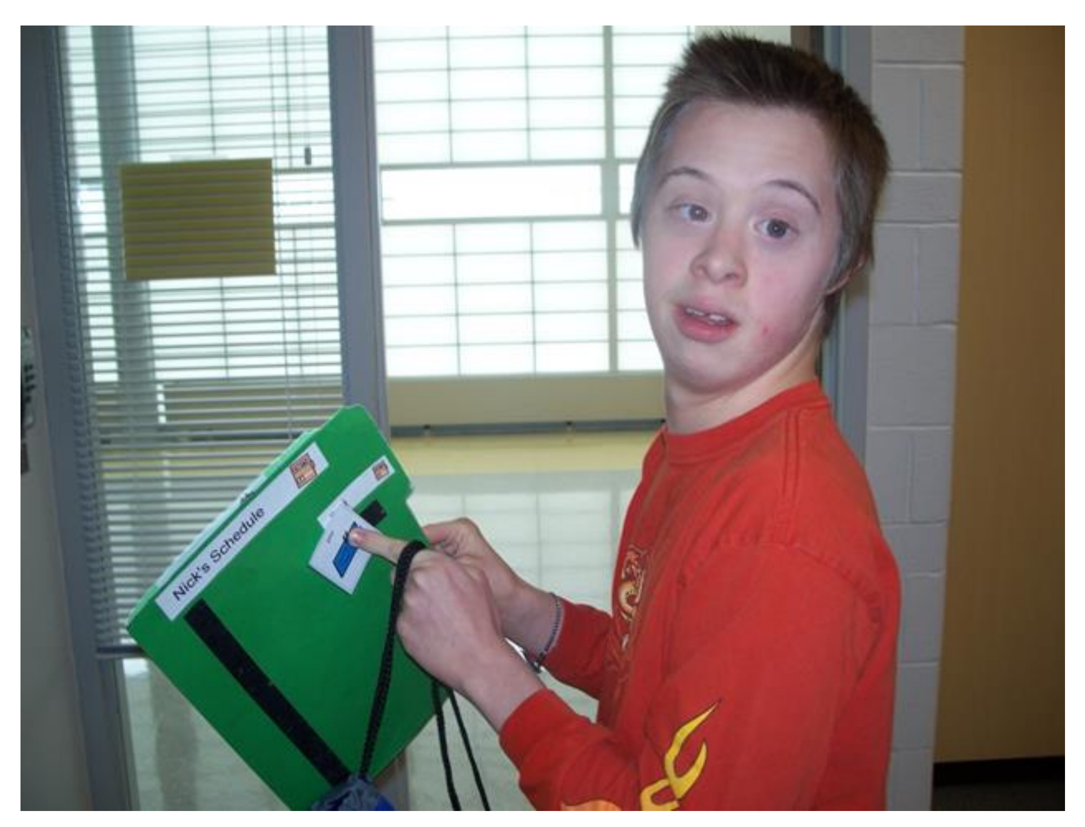

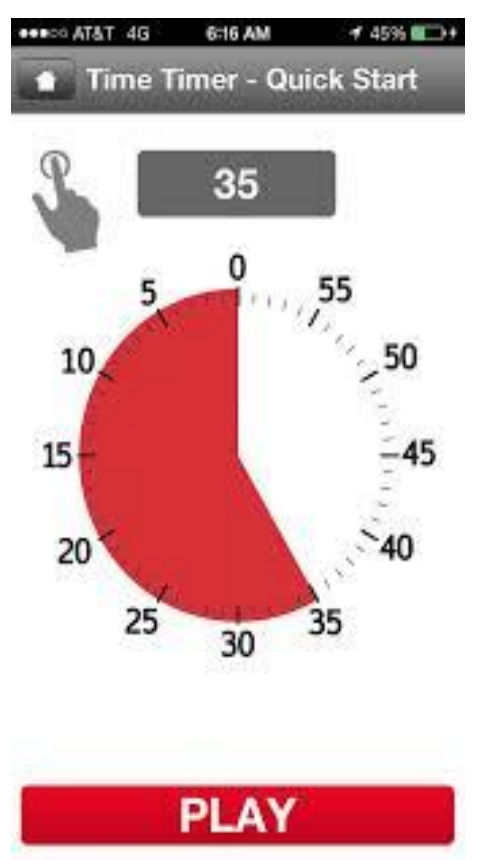

Behavior:
ABA Data Collection

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ABC FUNCTIONAL ASSESSMENT CARD

Definition of the behaviour:

CHILD'S NAME:

GENERAL CONTEXT:

OBSERVER:	**DATE and TIME:**	
Antecedent: *What happened before?*	**Behaviour:** *What did you see or hear?*	**Consequence:** *What followed?*

Fill this section later: Circle the function(s) demonstrated by this behaviour:

ESCAPE/AVOID:	**Attention**	**Object/activity**	**Sensory stimulation**
or			
OBTAIN/ GAIN ACCESS TO:	**Attention**	**Object/activity**	**Sensory stimulation**

OT and Sensory Diets

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Educational advocacy for co-occurring DS-ASD

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- Seek parent support for home and suggest specific DS-ASD support groups.
- Collaborate as a team at IEP meetings—look outside the box for solutions
- Determine what supports and accommodations are needed to make learning the most effective.
- Additional training for staff and aides—EX/ CPI, BSP reminders for para/aides.
- Safety concerns in the school building—Elopement, 1:1 para/aide
- Giving the student a voice– Ex/ IEP participation, need a break card with headphone pics, choice boards for activities, rewards, snacks.
- Lead with the student's strengths in the IEP plan.

Minimize Barriers → Maximize Strengths

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Nick's Strength's

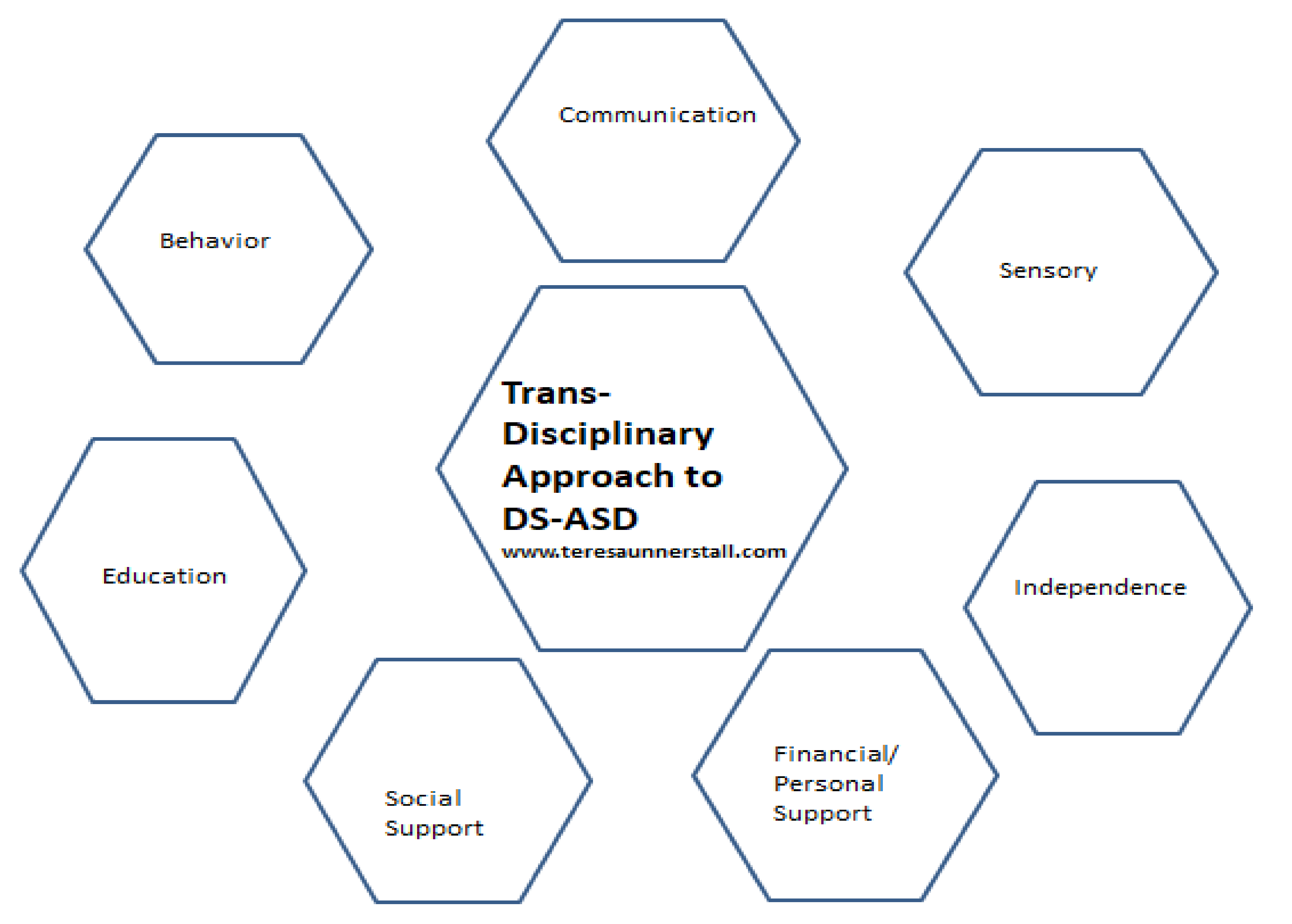
- Matching
- Object Permanence
- Desire to please
- Understands visuals
- Follows schedules
- Enjoys "heavy" work/activities
- Great sense of humor
- Socially engaging

Nick's Challenges

- Verbal perseverative play
- Stimming
- Impulsivity
- OCD tendencies
- Sensitive to criticism & correction
- Insistence on sameness
- Insistence on personal space (respect the bubble)

Home
Matching
Unload dishwasher
Recycling
Vacuuming

Community
Restore Packaging
Assisted living: vacuuming
GiGi's Playhouse – cleaning
Pet Shelter = sorting


Communication
Behavior
Sensory
Trans-Disciplinary Approach to DS-ASD
www.teresaunnerstall.com
Education
Independence
Social Support
Financial/ Personal Support

Nick’s Jobs

www.teresaunnerstall.com

Maintenance: Assisted Living Facility

www.teresaunnerstall.com

Never Set Limits!
Nick Diveheart Scuba Program

www.teresaunnerstall.com

Additional Supports:

www.teresaunnerstall.com for DS-ASD Consulting

- State Waiver for in home family support. SSI for qualifying families.
- Respite care/personal support workers
- Support groups specific to co-occurring DS-ASD- **The Down Syndrome-Autism Connection** www.ds-asd-connection.org (Facebook private group, Together Tuesdays)
- DSCBA Dual Diagnosis Support www.dscba.org
- Resource library, AAC templates and online links- www.dscba.org

Book available on Amazon and Blog, www.nickspecialneeds.com

Follow us on Facebook and Instagram at Down Syndrome With a Slice of Autism Access all the links at www.teresaunnerstall.com