

DUAL DIAGNOSIS
DOWN SYNDROME & AUTISM SPECTRUM DISORDER
Compiled by Dr. John Hartweger

INCIDENCE

Sources cite between 5-39% but 10% is probably most accurate
There is a much higher incidence in males

SIGNS AND SYMPTOMS

There are two groups of individuals with a dual diagnosis:

GROUP 1:

develop atypical behaviors early – infancy or toddler years
may have associated medical conditions such as seizures and hypotonia
repetitive motor behaviors; fascination with lights, fans, fingers; extreme food refusal; receptive language problems;
expressive language is repetitive or absent

GROUP 2:

usually older and experience a dramatic loss in their acquisition and use of language and social-attending skills
may be followed by excessive irritability, anxiety, and onset of repetitive behaviors
regression occurs between ages of 3-7 years

*SIGNS AND SYMPTOMS VARY – “SPECTRUM”

MOST COMMONLY DESCRIBED AREAS OF CONCERN WITH ASD

1. COMMUNICATION

Expressive language limitations; nonverbal
Receptive language limitations as well

2. SOCIAL SKILLS

Significant lack of social response or relatedness with family and friends
Definite lack of interest or ability in developing relationships with peers
Antisocial, anxious, fearful in the presence of people they don't know

3. REPETITIVE BODY MOVEMENTS OR BEHAVIOR PATTERNS

Hand flapping, spinning, rocking, shaking
Obsession with strings, lights, fans, mirrors, hands, fingers, water
No creative play; line up toys

SOME OF THE VARIABLE CHARACTERISTICS OF ASD IN CHILDREN WITH DOWN SYNDROME

1. Unusual response to sensations, especially sounds, lights, touch
2. Food refusal
3. Unusual play with toys and other objects – **repetitive movements or shaking of toys or objects**
4. Difficulty with changes in routine or familiar surroundings – **upset with changes in routine**
5. Little or no meaningful communication
6. Disruptive behaviors including aggression, tantrums, extreme non-compliance
7. Hyperactivity, short attention, impulsivity
8. Self-injurious behavior
9. Sleep disturbances
10. h/o development regression, especially language and social skills; **lack of eye contact, prefers to be left alone**

EVALUATION

*IN 2007, AAP RECOMMENDED ROUTINE SCREENING OF PATIENTS WITH DS FOR ASD AT 18-30 MONTHS

**** AN ACCURATE SCREENING TOOL IS NOT UNIVERSALLY RECOGNIZED FOR THIS HIGH-RISK POPULATION**

***** DELAYS IN DIAGNOSIS AND LACK OF RECOGNITION OF DEVELOPMENTAL REGRESSION HAVE AFFECTED OUTCOMES OF PATIENTS WITH DUAL DIAGNOSIS IN A NEGATIVE WAY**

TEAM

1. PHYSICIAN (NEUROLOGIST, PSYCHIATRIST, OR DEVELOPMENTAL PEDIATRICIAN)
2. SPEECH THERAPIST
3. PSYCHOLOGIST

QUESTIONNAIRES

1. AUTISM DIAGNOSTIC OBSERVATION SCHEDULE (ADOS)
2. AUTISM DIAGNOSTIC INTERVIEW – REVISED
3. MODIFIED CHECKLIST FOR AUTISM IN TODDLERS (M-CHAT)

MEDICAL WORK-UP

- A. A complete medical history: prenatal, neonatal information, developmental history, family history, medical history, educational history, interventions
- B. A complete medical evaluation
 1. HEARING EVALUATION
 2. VISION EVALUATION
 3. LEAD TEST
 4. CBC
 5. IRON STUDIES
 6. LFTS
 7. EEG
 8. SLEEP STUDY BY 4 YEARS OF AGE *The incidence of obstructive sleep apnea in people with Down syndrome is 50-70%*
- C. A psychological evaluation to assess cognitive functioning, adaptive behaviors, and presence of autism
- D. Speech and language evaluation to assess communicative intent and ability
- E. Occupational therapy evaluation – to assess sensory and motor problems typically associated with autism
- F. Psychosocial evaluation to assess the individual's home and school environment.

OBSTACLES TO DIAGNOSIS DS-ASD

1. Failure of parents to recognize, understand, agree or accept the diagnosis
2. Failure of therapists and health professionals to recognize or accept
 - a. The physician needs to recognize the child's loss of social skills, lack of progress in language, or lack of socialization
 - b. Many parents feel professionals had been dismissive of their concerns
 - c. Children with Ds will fail items testing language on autism screening BUT will typically pass items dealing with socialization, play and joint attention
3. Diagnostic overshadowing – Down syndrome interferes with detection
4. Diagnostic limitations

BEHAVIORAL FINDINGS

1. History of developmental regression including loss of language and social skills
2. Poor communication skills
3. Self-injurious and disruptive behaviors such as skin picking, biting, head hitting, head banging
4. Repetitive motor behaviors such as grinding teeth, hand flapping, rocking
5. Unusual vocalizations including grunting, humming, throat noises
6. Unusual sensory responsiveness such as spinning, staring at lights or sensitivity to certain sounds
7. Feeding problems such as food refusal or strong preference for specific textures
8. Increased anxiety, irritability, difficulty with transitions, hyperactivity, attention problems, sleep disturbances
9. Scored significantly higher than peers on 5 subscales of ABC: sensory function, social relating, body/object use, language use, social skills

10. Show less impairment in social relatedness than those with ASD only
11. Show more preoccupation with body movement and object use than children with ASD only
12. Scored higher than children with severe cognitive impairment on 5 subscales of ABC

ASSOCIATED MEDICAL CONDITIONS

DS/ASD children were more likely to have:

Heart disease and gi tract abnormalities, neuro findings including seizures, hypotonia, optho problems, respiratory problems

TREATMENT

“Consider the autism as the primary disability. Children may have Ds but it’s the autism that gets in the way of their learning and reacting in a way that you would expect.”

TARGET BEHAVIORS

Hyperactivity and poor attention
Irritability and anxiety
Sleep disturbance
Explosive behaviors
Rituals and repetitive behaviors
Self-injury

LANGUAGE THERAPY

STRATEGIES

Adequate preparation time for transitions
Offering visual and auditory cues before transitions
Singing familiar songs to decrease anxiety in difficult situations
Select settings with fewer potential triggers
Inclusion – in and out of school – “power of peers”

SOCIAL SKILLS

SPEECH THERAPY

OTHER CONSIDERATIONS

SOCIAL ISOLATION

“Today I know the isolation of being a mother of a child who is different from his peers with Down syndrome”

“Unfortunately, I have found that parents in this situation almost universally withdraw from local Down syndrome support groups...for a variety of reasons...”

“Topics discussed don’t apply to my child”

“It’s just too hard to see all those children doing so much more than my child”

“I feel like people think I’m a bad parent because of my daughter’s behavior”

ADJUSTING TO THE DIAGNOSIS

“But I was just getting used to Holland”

Sorrow, depression, fear, anger, frustration, resentment, jealousy, relief, sadness, guilt, disappointment, isolation, loneliness

Mother-Father relationship; parent-child relationship, family-extended family relationship, family-friends relationship

PREPARE A MEDICAL “RESUME”

Page 1 – Child’s name, address, parents’ names, phone numbers, insurance info

Page 2 – Diagnoses, medications, allergies, surgeries

Page 3 – Doctors’ names, addresses, phone numbers, fax numbers

Page 4 – Lab and test results

Page 5 – Hospitalizations, when and where

General Anesthesia

Dental work, deep cleanings, x-rays

Blood tests

Eye screenings

Hearing screenings

Immunizations

Gyn exams

X-rays, US, MRI, CT

RESOURCES

Autism Society of America - <http://www.autism-society.org/>

Autism Speaks - <http://www.autismspeaks.org/>

Disability Solutions Down Syndrome – Autism Connection - <http://www.ds-asd-connection.org/>

Kennedy Krieger Institute - https://www.kennedykrieger.org/patient-care/outpatient-programs/dual_diagnosis

National Association for Dual Diagnosis (NADD) – <http://thenadd.org/>

National Down Syndrome Society (NDSS) – <http://www.ndss.org/Resources/Health-Care/Associated-Conditions/Dual-Diagnosis-of-Down-Syndrome--Autism/>

When Down Syndrome and Autism Intersect: A Guide to DS-ASD for Parents and Professionals, Foehlke, Margaret, RN (2013)

DSAGSL Website - <http://dsagsl.org/programs-resources/resources/dual-diagnosis>