

DIAGNOSIS, TREATMENT AND LONG-TERM CONSIDERATIONS FOR THE AUTISTIC INDIVIDUAL:

FOUNDATION FOR THE LIFE CARE PLAN

KRISTI B. BAGNELL, MD, FAAP, CLCP
 NANCY J. BOND, CCM, CLCP

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OBJECTIVES:

- ▶ Describe the screening and diagnosis of Autism Spectrum Disorder (ASD)
- ▶ Discuss causation of ASD: genetic and environmental associations
- ▶ Review associated medical and mental health conditions of ASD
- ▶ Describe therapeutic behavioral treatment of ASD (ABA therapy)
- ▶ Address other current and future treatments of ASD
- ▶ Discuss challenges of transition to adult care for autistic persons
- ▶ Case Reports

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AUTISM SPECTRUM DISORDER DEFINITION AND HISTORY

- ▶ "Complex developmental condition that involves persistent challenges in social interaction, speech and nonverbal communication and restricted/repetitive behaviors" (American Psychiatric Association, 2019)
- ▶ First described by Leo Kanner, MD in 1943 - "Powerful desire for aloneness"
- ▶ 1980 - Recognized as a diagnosis in *DSM-III*
- ▶ 2000 - Deficits in 3 domains required in *DSM-IV TR*
- ▶ 2013 - Prior categories absorbed into new diagnosis of Autism Spectrum Disorder for *DSM-5*

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AUTISM SPECTRUM DISORDER DIAGNOSIS

- ▶ Currently no reliable biomarkers to identify ASD
- ▶ Diagnostic tools for ASD: Autism Diagnosis Interview - Revised (ADI-R), Autism Diagnostic Observation Schedule Generic (ADOS-G), Childhood Autism Rating Scale (CARS), Gilliam Autism Rating Scale - Second Edition (GARS-2)
- ▶ Diagnostic tools used with DSM-5 standardized diagnostic criteria
- ▶ Diagnostic criteria are categorized according to deficits in the two domains of social communication and interactions across multiple situations (currently or by history) **AND** restricted interests and repetitive behaviors

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AUTISM DIAGNOSIS

- ▶ The restricted or repetitive patterns must be manifested by certain characteristics:
 - ▶ Stereotyped or repetitive speech or motor movements, inflexible adherence to routines, highly restricted and fixated interests that are abnormal in intensity, and hyper- or hypo-reactivity to sensory input
"Cade"
- ▶ Each category must also specify a **Severity Level** of function



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AUTISM SEVERITY LEVELS

- ▶ **ASD Level 1:** Requiring Support (Difficulty initiating social interactions, organization and planning problems can hamper independence)
"Hannah"
- ▶ **ASD Level 2:** Requiring Substantial Support (Social interaction limited to narrow special interests, frequent restricted and/or repetitive behaviors)
- ▶ **ASD Level 3:** Requiring Very Substantial Support (Severe deficits in verbal and nonverbal social communication skills, great distress and/or difficulty changing actions or focus)



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AUTISM SPECTRUM DISORDER CAUSATION

- ▶ Genetic factors contribute to the majority of ASD
- ▶ Other risk factors have been identified:
 - ▶ Advanced parental age (Mother > 35, Father > 40)
 - ▶ Second child affected with autism > 18%
 - ▶ Non-identical twins, other twin affected 31%
 - ▶ Identical twins, other twin affected 50-70%
- ▶ Autism spectrum disorder results from a combination of genetic and environmental influences (Hormozdiari, et al, 2018)

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AUTISM SPECTRUM DISORDER CAUSATION

- ▶ Identified areas in the brains of those affected by autism
 - ▶ Postmortem studies: limbic system, cerebellum
 - ▶ Functional MRI: areas responsible for social and affective judgements, areas that process facial and non-facial motives
 - ▶ Structural MRI: increased overall size of brain
 - ▶ DTI: defective development of white matter tracts
- ▶ Elevation in serotonin levels, dysregulation of dopamine and branched-chain amino acids

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AUTISM SPECTRUM DISORDER CAUSATION

- ▶ Single-gene syndromes: Fragile X (1-3% of ASD cases)
- ▶ Hundreds of genes contribute to ASD
 - ▶ Copy number variations testing identifies genetic mutations: deletions or duplications
- ▶ Epigenetics: what turns genes "off and on"
 - ▶ Research lags significantly behind mutation studies
 - ▶ Methylation testing: reveals methylation alterations in distinct genetic disorders and environmental exposures

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AUTISM SPECTRUM DISORDER CAUSATION

- ▶ Measles-Mumps-Rubella vaccine and thimerosal-containing preservative: no scientific evidence to support as causative agents
- ▶ Possible role of immune system in some cases of autism (Pardo, et al, 2005)
- ▶ Abnormalities in the immune system in ASD: increase in activated B cells, natural killer cells, and pro-inflammatory cytokines; dysfunction of T cells and autoantibody production

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AUTISM SPECTRUM DISORDER ASSOCIATIONS

- ▶ Meta-analysis of the association between perinatal and neonatal factors and autism risk (Gardener, et al, 2011)
- ▶ Abnormal presentation, umbilical cord complications, fetal distress, birth injury or trauma, multiple births, maternal hemorrhage, summer birth, low birth weight, small for gestational age, congenital malformation, low Apgar score, feeding difficulties, meconium aspiration, neonatal anemia, ABO or Rh incompatibility, hyperbilirubinemia
- ▶ "Etiologically relevant period" extends from in utero through early infancy

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AUTISM SPECTRUM DISORDER MEDICAL TREATMENT CONSIDERATIONS

- ▶ Epilepsy affects 20-30 % persons with ASD. Intellectual disability further increases the risk for developing clinical seizures. Up to 60% with ASD have abnormal EEGs.
- ▶ Sleep disorders in up to 80% of children with ASD
- ▶ Gastrointestinal abnormalities: 8 times more likely in children with ASD: constipation, GERD, food allergies, obesity in association with syndromes, medications or lack of exercise
- ▶ Dental problems and decay

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AUTISM SPECTRUM DISORDER MEDICAL TREATMENT CONSIDERATIONS

- ▶ Behavioral abnormalities are commonly observed in ASD:
 - ▶ Attentional deficits and hyperactivity, aggression, self-injurious behaviors, obsessive-compulsive symptoms, stereotypies, tics, lability, anxiety, depression
 - ▶ Currently, risperidone and aripiprazole are the only medications approved by the FDA for irritability, physical aggression and severe tantrum behavior associated with ASD
 - ▶ Goal of pharmacological treatment: improve the benefit from behavioral treatments and educational activities while remaining in a less restrictive environment (Volkmar, et al, 2014)

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AUTISM SPECTRUM DISORDER MEDICAL TREATMENT CONSIDERATIONS

- ▶ Attention and hyperactivity disorders in 30-61% of ASD
 - ▶ 49% had positive response to treatment with methylphenidate in large randomized controlled trial (Research Units on Pediatric Psychopharmacology Autism Network, 2005)
- ▶ Anxiety disorder in 11-40% children and teens with ASD
- ▶ Schizophrenia in 4-35% of adults with ASD
- ▶ Females with ASD have 3 times risk of suicide, young individuals with ASD with twice the risk of suicide (Kirby, et al, 2019)

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AUTISM SPECTRUM DISORDER MEDICAL TREATMENT CONSIDERATIONS

- ▶ Unfortunately, there is a lack of evidence for most forms of psychological intervention in ASD
- ▶ Exception: cognitive behavioral therapy for anxiety and anger management in high functioning youths with ASD
- ▶ Hospitalized in psychiatric units a much higher rates than children without ASD
- ▶ Efficacy only if the facility specializes in autism treatment (Volkmar, et al, 2014)

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AUTISM SPECTRUM DISORDER CHALLENGES

- ▶ One-third of autistic persons are non-verbal (many benefit from augmentative communication devices)
- ▶ 28% of 8-year-olds with ASD displayed self-injurious behaviors (Soke, et al, 2016)
- ▶ Many with autism have a unique fascination with water or other hazardous areas (active roadways or around trains)
- ▶ Nearly half with ASD wander or bolt from safety
- ▶ Drowning is the leading cause of death for children age 14 and younger with ASD, accounting for 90% of deaths associated with wandering or bolting

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AUTISM SPECTRUM DISORDER BEHAVIORAL TREATMENT CONSIDERATIONS


- ▶ Applied behavior analysis (ABA) has evolved as an effective treatment for ASD
- ▶ Uses the principles of behavior to produce meaningful change
- ▶ ABA procedures help increase adaptive skills, social behavior and self-help skills; decrease stereotypy and tantrums
- ▶ Published studies note the importance of beginning treatment prior to 3 years of age, with the most significant gains when treatment is initiated before 24 months of age (MacDonald, et al, 2018)

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AUTISM BEHAVIORAL THERAPY

- ▶ Applied Behavioral Therapy (ABA) techniques:
 - ▶ Pivotal Response Treatment
 - ▶ Early Start Denver Model
 - ▶ Verbal Behavior Therapy
- ▶ Discrete trial instructions: primarily provider-directed, teaching in simplified and structured steps "Cub"
- ▶ Naturalistic instructions: primarily client-directed, providing choices to increase motivation to learn



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AUTISM SPECTRUM DISORDER BEHAVIORAL TREATMENT CONSIDERATIONS

- ▶ Behavioral Analyst Certification Board published guidelines for ABA treatment in 2014
- ▶ Intensity of treatment should range from 10-25 hours/week for focused treatment, and 30-40 hours/week of 1:1 comprehensive treatment
- ▶ Focused ABA treatment may include more than 25 hours/week in day treatment or inpatient program for severe self-injurious behavior
- ▶ ABA has been shown to be effective across the life span
- ▶ New CPT coding for ABA therapy went into effect on January 1, 2019 (Association for Behavioral Analysis International 2018)

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AUTISM SPECTRUM DISORDER OTHER TREATMENT CONSIDERATIONS

- ▶ Occupational therapy
- ▶ Speech/language therapy
 - ▶ Alternative augmentative communication (AAC)
- ▶ Psychological assessment
- ▶ Structured educational plan with explicit teaching
 - ▶ Treatment and Education of Autism and related Communication Handicapped Children program (TEACCH)

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AUTISM SPECTRUM DISORDER COMPLIMENTARY, INTEGRATIVE TREATMENT

- ▶ Biomedical Therapies: Melatonin, omega-3 fatty acids, injectable methylcobalamin (methyl B12), N-acetylcysteine, pancreatic digestive enzymes, probiotics, micronutrients, and immune therapies (IVIG) (Hendren, et al, 2018)
- ▶ Music Therapy
- ▶ Art Therapy
- ▶ Animal Therapy

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AUTISM SPECTRUM DISORDER TREATMENTS UNDER INVESTIGATION

- ▶ Repetitive transcranial magnetic stimulation
 - ▶ Valid treatment in a small number of trials
 - ▶ Improvements in neuropsychological performance and reduction in core symptoms and associated features of ASD (Enticott, 2018)
- ▶ Stem cell therapy
 - ▶ Duke University clinical trial used autologous umbilical cord blood in 25 children: significant improvements in social communication skills and autistic symptoms by parental report; overall improvements in autism symptom severity by clinician ratings; better expressive vocabulary and objective eye-tracking of attention to social stimuli by standardized measures (Dawson, et al, 2017)

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AUTISM SPECTRUM DISORDER LONG TERM CARE PLANNING CONSIDERATIONS

- ▶ Estimated 50,000 teens per year with autism will age out of school-based services over the next decade
- ▶ Teens with autism have been estimated to receive transition services half as often as others with special healthcare needs, even less if with associated medical disorders (Cheak-Zamora, et al, 2013)
- ▶ More than half of young adults with ASD are unemployed in the two years after high school (lower than young adults in other disability categories)
- ▶ 60% who attended state-funded vocational programs attained employment, 80% of which were part-time jobs (Chen, et al, 2015)

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AUTISM SPECTRUM DISORDER LONG TERM CARE PLANNING CONSIDERATIONS

- ▶ An "autism epidemic" is rapidly approaching adulthood
- ▶ Estimated that a majority of those with ASD will require some type of residential service or support
- ▶ Residential services in extremely short supply for adults with ASD
- ▶ Exceptional programs: difficult to find, lack availability
- ▶ Vendor surveys present a challenge for life care planners

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AUTISM SPECTRUM DISORDER COST OF CARE

- ▶ Estimated annual cost of caring for Americans with autism is \$268 billion. Without more effective interventions and support, the predicted cost will rise to \$461 billion by 2025 (Leigh, et al, 2015)
- ▶ Joint US and UK study found that ASD is the fourth most expensive condition in both countries, behind trauma, cancer and CV disease (Buescher, et al, 2014)
 - ▶ For children, highest costs were related to special education services and parental productivity loss
 - ▶ For adults, highest costs were related to residential care or supportive living accommodations and individual productivity loss
 - ▶ Medical costs were substantially higher for adults than for children

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AUTISM SPECTRUM DISORDER LEGISLATION

- ▶ Combating Autism Act: signed into law in 2006 as a coordinated federal response to the dramatically rising numbers of individuals diagnosed with ASD
- ▶ Reauthorized in 2014 as the Autism Collaboration, Accountability, Research, Education, and Support (CARES) Act: bipartisan bill, passed unanimously in House and Senate, focused on transition services for students
- ▶ Reauthorization of CARES occurred again on September 30, 2019: will focus on ASD across the lifespan, including better health care, education, employment, and home and community-based services and supports

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CONCLUSION

- ▶ Due to the heterogeneity of the effects and comorbidities for individuals on the autism spectrum, life care planning is and will be challenging.
- ▶ The results of diagnostic testing for ASD will be useful in the development of the life care plan, as specifiers identify the severity of symptoms and comorbid conditions.
- ▶ Life care planners must be aware of all the autistic individual's medical and mental health disorders to adequately project future diagnostic, treatment, and long-term care needs.
- ▶ ABA therapy has emerged as an effective behavioral intervention for ASD and should be instituted as soon as possible for best outcomes.

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CONCLUSION

- ▶ Occupational and speech therapy are often adjuvants to therapeutic treatment.
- ▶ Augmentative communication devices are helpful for many non-verbal individuals with autism.
- ▶ Music therapy, art therapy and animal therapy, as well as complementary biomedical treatments should be considered on an individual basis.
- ▶ Promising treatments for ASD are currently being investigated.
- ▶ Future genetic and immunological research efforts using diagnostic biomarkers, methylation alterations and immune dysfunction hope to reveal targeted therapeutic interventions.

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CONCLUSION

- ▶ Perhaps the most difficult projection for those on the autism spectrum will be for their long-term care, including provisions for education, employment, residential care and other community-based activities.
- ▶ A life care plan for the person with autism spectrum disorder will undoubtedly enhance their quality of care and serve as a basis for financial planning to ensure a safe, healthy and productive life.

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AUTISM SPECTRUM DISORDER AND LIFE CARE PLANNING

Case Study #1

"Mary"

- 4 years old at time of referral for life care plan
- Born full term by repeat C-section
- Complications at birth
- At 9 month check-up, pediatrician noted delay in acquisition of skills
- Developmental evaluation revealed moderate gross motor delay, mild fine motor delay and significant delays in cognitive and adaptive skills; no delays in language skills
- At two years of age, she was finally able to stand and take a few steps
- At 30 months of age, still with significant delays in cognitive/adaptive and language skills

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AUTISM SPECTRUM DISORDER AND LIFE CARE PLANNING

Mary's Life Care Plan

- Observations of child at both home and at school
- Consultation/input provided by both treating and expert specialists
- Projected services:
 - Developmental pediatrics throughout growth years
 - Ongoing surveillance by neurology and PM&R services
 - Behavioral intervention/supports for parents
 - Incontinence supplies through age 8; anticipate continence would be achieved with reminders/scheduling
- Equipment
- Rehabilitation therapies
- Home care
- Transportation to and from therapy sessions

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AUTISM SPECTRUM DISORDER AND LIFE CARE PLANNING

Case Study #2

"Larry"

- 19 years old at time of referral for life care plan
- Born full term via STAT C-section due to bradycardia and prolapsed cord
- Apgar scores 1, 4 and 5 at 1, 5 and 10 minutes respectively
- Returned to hospital at 8 days of age with apnea and cyanosis
- Early pediatric history significant
- Seizure disorder diagnosed at 15 years of age (tonic-clonic seizures)

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AUTISM SPECTRUM DISORDER AND LIFE CARE PLANNING

Larry's Life Care Plan

- Interview of mother at office and then observations of Larry at school
 - Mother reported inability to control Larry within the home
 - School provided insight into behavior in more challenging situation
- Consultation/input provided by expert specialists (mother had not taken Larry to physician other than PCP for some time)
- Projected services:
 - Neurology and PM&R
 - Behavioral intervention/supports for mother
 - Equipment
 - Rehabilitation therapies
 - Home care
 - Transportation to and from therapy sessions
