

Medical Causes of Maladaptive Behavior in ASD

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Objectives

- To give tools to assess autistic individuals for medical problems that could present as maladaptive behaviors
- To provide participants with the means to identify when to suggest a referral and who is the appropriate HCP
- To illustrate potential dysfunctions that have been linked to autism and comorbid medical disease in the peer reviewed literature

Introduction...



Medical or Behavioral?

- Why is she/he acting this way?
 - People with autism require a specialized medical approach to diagnose underlying disease
 - **DO NOT DISMISS BEHAVIORS AS “JUST THE AUTISM”**
- Anyone with training in behavior analysis knows that ruling out a physiological function of behavior is the first step when doing a FBA
 - **THIS IS EASIER SAID THAN DONE!**

Why?

“ Almost all patients referred for a GI work-up have a normal physical exam. In fact, a *normal physical exam* should not exclude GI issues or a referral to a gastroenterologist. Behaviors particularly aggression and self injury **are** the symptoms that should motivate the HCP to look further.”

- Timothy Buie, M.D.
Lurie Center for Autism
Massachusetts General Hospital for Children
Gastrointestinal Comorbidities in Autism Spectrum Disorders

Link to webcast: www.ccfme.org/autism15

Autism Centers of Excellence

- ACE Program is an NIH initiative that supports large scale studies that are research focused:
 - Boston University
 - Emory University
 - University of California, Los Angeles
- Centers that focus on coordinating medical care across multiple specializations:
 - Massachusetts General Hospital (Lurie Center for Autism)
 - Cleveland Clinic Center for Autism
 - University of California, Davis
 - And more...

Treatment Oriented Centers

- Coordination of care and expertise in:
 - Neurology
 - Developmental Pediatric Medicine
 - Gastroenterology
 - Psychiatry / Psychology
 - Genetics
 - Metabolic
 - Nutrition
 - Allergy / Immunology
 - Special Needs Dentistry
 - OT, PT, Speech Therapy
 - Nurse Case Management
- Focus on lifelong care



Good Assessment is Vital

- Children & Adults with Autism
 - 50% are non-verbal or minimally verbal
 - Most have sensory issues
 - Affects ability to communicate symptoms effectively
- Proprioceptive issues
 - Body is lost in space
 - Can not identify or point to what is hurting
 - Delays in motor planning and fine motor skills



Barriers to Diagnosis of Treatable Medical Conditions

- Communication Impairments
- Social Impairments (muted cues & lack of imitation skills)
- Sensory & Motor Impairments
- High Pain Tolerance in some individuals
- Knowledge deficits on the part of health care providers (HCP)



- HCP especially Emergency Response Teams and ER staff:
- During a behavioral crisis
 - Often a medical exam NOT done in the emergency room
 - Assumed to be psychiatric - "Baker Act"
 - Patient is not worked up for medical or neurological contributing factors
- Assumption of staff who work with autistic individuals
 - That changes in behavior are (solely) behavioral
 - CONSIDER other possible factors such as pain, inflammation, infection, etc.



Medical Detective



Assessment Tools

- Ask the Parents or Caregivers
 - Understand baseline behaviors
 - Important to detect changes even if subtle
- DO NOT pre judge on outward behaviors & labels
- Stims (Self Stimulatory Behaviors)
 - Can be a clue on what bothering a person
 - Look at the stims when assessing behavior change
 - Some repetitive movements are an attempt to alleviate pain



Strategies for Communication

- Teach Pain Scale when not in pain
- Teach about the body and the way organs work
- Presume competence
- Use age appropriate language
- Use whatever educational methods that have been successful
 - DTT (Discrete Trial Training)
 - Social Story (check comprehension)
 - Visuals / Assistive Technology
 - RPM (teach/ask format)
 - Choice Boards



Choice Board

Physical	Emotional	Something Else
Pain	Seizure	Sad
Anxiety	Tired	Anger

Signs vs Symptoms?

- Assessment includes: **Signs** (objective) & **Symptoms** (Subjective)
- Both are evidence of disease
 - Baseline is important
 - Unique S&S specific to the individual
 - A medical issue can turn into a behavior problem
 - A behavioral problem or stim can lead to a medical problem
 - Treat both for best outcome
 - Sudden onset or change in behavior
 - Changes always call for a complete medical workup



Potential Problems...

- Gastrointestinal
- Neurological
- Immune
- Metabolic / Endocrine
- Urinary
- Mitochondrial Disorders
- Dental Issues



References



PEDIATRICS

OFFICIAL JOURNAL OF THE AMERICAN ACADEMY OF PEDIATRICS

Recommendations for Evaluation and Treatment of Common Gastrointestinal Problems in Children With ASDs
 Timothy Bui, George J. Dunbar III, Glenn T. Fennie, Kenneth Krasner, Joseph Leroy, Jeffrey D. Lewis, Barry K. Wondol and Hetalal Water
Pediatrics 2016;137:515-519
 DOI: 10.1542/peds.2016-1876D

The online version of this article, along with updated information and services, is located in the World Wide Web at www.pediatrics.com/cgi/content/full/137/Supplement_1/515-519

PEDIATRICS

OFFICIAL JOURNAL OF THE AMERICAN ACADEMY OF PEDIATRICS

Evaluation, Diagnosis, and Treatment of Gastrointestinal Disorders in Individuals With ASDs: A Consensus Report
 Timothy Bui, George J. Dunbar, George T. Fennie III, Glenn T. Fennie, Joseph Leroy, Jeff VanDerWalt, Agnes H. Winkler, Dan Adams, Margaret L. Berman, Andrew L. Daniels, Edward A. Cook, Michael D. Goren, Steven M. Hirsch, Pratik Kapadia, Harriet Kopyov, Kristina Kover, Shari Kurland, Pat Levin, Sarah E. Lyle, Jeffrey D. Lewis, Katherine J. Meyer, Marissa B. Nemerov, Akshat Natta, Barry K. Wondol, Sherry Y. Wang, Louise Zeman and Hetalal Water
Pediatrics 2016;137:515-519
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Take Home Points

- Problem Behaviors may be the **PRIMARY** symptom of underlying medical conditions including GI disorders
- Limited evidenced based studies to guide clinicians
- Recommendations to evaluate and treat based on expert opinions across multiple specializations
- Initiate behavioral treatment while investigating medical illness **BUT** behavioral therapy should not substitute for medical investigation



Prevalence of GI Issues

- Presentation by Dr. Buie, Pediatric Gastroenterologist
40-70% of the ASD population suffers from GI issues
- * 2014 Meta-analysis conducted by McElhanon, et. al.
50-70% of ASD population suffers from a GI disease or condition
- GI issues disproportionately higher than in the general population

Information taken from:
Gastrointestinal Comorbidities in ASD
Timothy Buie, M.D.
www.ccfcmc.org/autism15

- *Gastrointestinal symptoms in ASD: a meta-analysis*
www.pediatrics.aappublications.org

Gastrointestinal System

- Underlying issues specific to ASD
 - Low oral motor tone
 - Affects the ability to chew
 - Digestive issues
 - Sensory Issues
 - Pertaining to texture and smell of food
 - Lead to nutritional deficiencies
 - Self Avoidance of certain foods
 - Could be an allergy or food intolerance
 - Low muscle tone
 - Leads to issues with slow bowel motility

* Refer to nutritionist and/or feeding specialist

GI Disorders

- Abdominal Pain
- Reflux / GER or GERD
- Esophagitis
- Constipation
- Diarrhea
- Malabsorption / Maldigestion
- Food Allergy or Food Intolerance
- Bloating and Gas
- Ulcers / Inflammatory Bowel Disease
- SIBO
- Celiac Disease

GI Issues in ASD

- Challenging to assess especially abdominal pain
- Atypical presentations of common GI problems
- Behaviors that could indicate GI pain:

Screaming
Posturing
Crying
Agitation
Irritability

Facial Grimacing
Jumping up and down
Self Injury
Aggression
Sleep Disturbances

* Refer to Gastroenterologist

Signs of Reflux / GERD

- Bending over
- Burping
- Throat clearing
- Guttural vocalizations
- Dry cough or habit cough
- Rumination
- Vomiting
- Crying & screaming especially after eating
- Sleep issues especially unwillingness to lay flat
- Change in eating habits
- Eating often & craving carbs

Symptoms of Reflux / GERD

- Burning in throat or chest
- Pain (Highly individualized as to location)
- A sour or bitter taste in the mouth
- Example: "Adams Apple Pain"

Signs of Constipation

- Straining & stools that are hard or small in diameter
- Diarrhea (overflow)
- Staining on underwear "encopresis"
- Rectal digging behavior
- Pointing or tapping on abdomen
- Posturing and putting pressure on the abdomen
- Distended abdomen



Symptoms of Constipation

- Abdominal pain
- A sense that everything didn't come out
- Complaints of nausea
- Complaints of uncomfortable abdomen
- Fatigue and feeling tired
- Appetite changes



Diarrhea

- Can be accompanied with or without abdominal pain
- Extremely soft or watery stools
- Frequent stools (greater than 3x per day)
- Undigested food in the stool
- Foul smelling stools
- Abdominal distention / Gas
- Can be associated with nutritional deficiencies



Food Intolerance or Sensitivities

- Difficult to detect by standard blood or skin tests
- Not IgE mediated
- Undigested food in stool
- Diarrhea & foul smell
- Eczema
- Non celiac gluten intolerance
- Salicylate / Oxalate issues



* Refer to Nutritionist experienced in ASD

Neurological Disorders

Most common in people with autism

- Epilepsy
- Sleep disorders
- Migraine headaches



Epilepsy

- Presentation by Dr. Bauman, Pediatric Neurologist
 - Seizures will occur in 1 out of 3 individuals with autism
 - Not one seizure type is prevalent
 - High risk periods
 - 0 – 5 years old
 - Adolescents
 - Young Adults

Information taken from:
Neurological Comorbidities in ASD
 Margaret Bauman, M.D.
www.cfcme.org/autism15



Epilepsy

- Subclinical epilepsy or abnormal EEG (spikes)
 - Non clinical seizures
 - Undo learning that takes place during the day
 - Can happen when sleeping
 - May not be easily recognized (staring off for a few seconds)
 - May play a role in psychiatric symptoms and behavioral disturbances
- Refer to a neurologist for 24 hour EEG and MRI if indicate



Reference

Frontiers in
PUBLIC HEALTH

REVIEW ARTICLE
published: 13 September 2015
doi: 10.3389/fpubh.2015.00011

A review of traditional and novel treatments for seizures in autism spectrum disorder: findings from a systematic review and expert panel

Richard E. Frye^{1*}, Daniel Rossignol^{2†}, Manuel F. Casanova³, Gregory L. Brown⁴, Victoria Martini⁵, Stephen Edelson⁶, Robert Cohen⁷, Jeffrey Levine⁸, John C. Slattery⁹, Chrystal Lau¹⁰, Paul Hardy¹¹, S. Hossain Fatemi¹², Timothy D. Folsom¹³, Derrick MacFabe¹⁴ and James B. Adams¹⁵

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⁸ Florida Healthcare Associates, Highland, MA, USA
⁹ University of Minnesota Medical School, Minneapolis, MN, USA
¹⁰ University of Western Ontario, London, ON, Canada
¹¹ Arizona State University, Tempe, AZ, USA

http://www.amazingbrains.com/wp-content/uploads/2014/05/article_file_4seizures-in-ASD.pdf



Migraine

- Head pain may present as self injury
- Observe for light and sound sensitivity with symptoms of headache or self injury
- May be difficult to diagnose if neurologist is not familiar with autism challenges
- Steroid, NSAID or inotropic trial may be indicated as a diagnostic tool



Sleep Disturbances

- Sleep Onset
- Sustaining Sleep
- Night Terrors
- Sleep Walking
- Restless Legs

*Refer to Sleep Specialist



Sleep Disturbances

- Possible Causes
 - Reflux
 - Seizures
 - Enlarged tonsils or adenoids
 - Anxiety and other psychological causes
- Sleep study including REM assessment may be indicated after 24 hour EEG evaluation



Immunological Issues

- Frequent illness and infections:
 - Otitis Media
 - Sinusitis
 - Pharyngitis
 - Bronchitis / Pneumonia
 - Recurrent strep
 - Chronic viral, bacterial, and/or yeast infections
 - Immune Deficiency (hypogammaglobulinemia)
- Frequent antibiotics in children can lead to long term problems
- Autoimmunity

* Consult PCP for referral to ENT or Immunologist



Immunological Disorders

- Allergies IgE mediated (food or environmental)
- Eczema and other skin conditions
- Enlarged tonsils and adenoids
- Allergic shiners
- Asthma

Testing is not 100% reliable

Studies show allergy prevalence higher in ASD population



* Refer to Allergist or Immunologist

Neuroimmunological Condition

- PANDAS (Pediatric Autoimmune Neuropsychiatric Disorder Associated with strep)
- PANS (Pediatric Acute Neuropsychiatric Syndrome)
- Both are defined by 5 criteria:
 - Abrupt, significant onset of OCD and/or Tics
 - Includes other neuropsychiatric symptoms
 - Prepubertal onset
 - Association with streptococcal or other infections
 - Symptoms following relapsing - remitting course
- Refer to a neurologist or immunologist for diagnosis & treatment



For Families and Support go to: www.pandasnetwork.org

For Practitioners go to: www.pandasppn.org

General information and advice: www.nimh.nih.gov

Reference

JOURNAL OF CHILD AND ADOLESCENT PSYCHOPHARMACOLOGY
Volume 24, Number 4, 2014
© Mary Ann Liebert, Inc.
Pg. 1-11
DOI: 10.1089/jcap.2014.0094

Consensus Statement

Clinical Evaluation of Youth with Pediatric Acute Onset Neuropsychiatric Syndrome (PANS): Recommendations from the 2013 PANS Consensus Conference

Kiki Chang, MD,^{1*} Jennifer Frankovich, MD,^{2*} Michael Cooperstock, MD, MPH,³
Madeline Cunningham, PhD,⁴ M. Elizabeth Laitner, MD,⁵ Tanya K. Murphy, MD,⁶
Mark Pasternack, MD,⁷ Margo Thiessenmann, MD,⁸ Kyle Williams, MD,⁹ Julian Walter, MD,¹⁰
and Susan E. Swedo, MD¹¹; From the PANS Collaborative Consortium

<http://www.ncbi.nlm.nih.gov/pmc/articles/PMC4340805/>



Urinary Issues

- Limited research as to how it applies to ASD
- Signs & Symptoms
 - Incontinence
 - Difficulty voiding
 - Frequency
 - Cloudy Urine
 - Constipation → Urinary Retention → Urinary Tract Infection
- Conditions
 - Spastic Bladder
 - Oxalate Issues
 - Recurrent bladder infection (can be associated with poor hygiene)



Metabolic / Endocrine Dysfunction

- Issues
 - Growth problems (refer to growth chart history)
 - Weight Gain (can be linked with behavioral meds)
 - Metabolic Syndrome
 - Thyroid and Adrenal issues
 - Hormone imbalances in both males and females
 - Rule out metabolic disease as a cause of ASD "inborn errors"



* Refer to appropriate specialist

Mitochondrial Disease

- Can occur at any age
- No difference between Mito disease or disorder (CDC)
- Mitochondrial disorders are commonly associated with epilepsy
- Treatment may optimize seizure management and improve developmental skills
- Research into mitochondrial dysfunction and ASD is ongoing



For more information:
Metabolic Comorbidities in ASD
Mark Korson, M.D.
www.ccfme.org/autism15

Bridging the Gap Between ASD and Mitochondrial disease
By Fran Kendall, M.D.
<http://www.mitoaction.org/files/AutismOne2011.pdf>

BRAIN	• Development delay	• Migraines	• Seizures
• Dementia	• Ataxia, balance	• Atypical cerebral palsy	
• Neuropsychiatric disturbances	• Mental retardation	• Strokes	
HEARS	• Absent reflexes	• Flaring	• Neurogenic pain
• Weakness (may be intermittent)	• Dysautonomia (e.g., temperature instability)		
MUSCLES	• Weakness	• Hypotonia	
• Cramping	• Muscle pain		
GASTROINTESTINAL	• Gastrointestinal problems	• Constipation/diarrhea	
• Intake bowel syndrome	• Diarrhea or constipation		
• Dysmotility			
• Pseudo-obstruction			
KIDNEYS	• Renal tubular acidosis or wasting		
HEART	• Cardiomyopathy	• Cardiac conduction defects (heart blocks)	
LIVER	• Liver failure	• Hypoglycemia (low blood sugar)	
EARS & EYES	• Vision loss and blindness	• Optic atrophy	• Acquired strabismus
• Ptosis	• Hearing loss and deafness	• Retinitis pigmentosa	
• Ophthalmoplegia			
PANCREAS & OTHER GLANDS	• Diabetes and excessive pancreatic failure (ability to make digestive enzymes)		
• Pancreatic failure (low calcium)			
SYSTEMIC	• Failure to gain weight	• Unexplained vomiting	• Respiratory problems
• Fatigue	• Short stature		

Bridging the Gap Between ASD and Mitochondrial disease
By Fran Kendall, M.D.
<http://www.mitoaction.org/files/AutismOne2011.pdf>

TIER 1 Basic workup recommended for all patients	TIER 2 Dependent on clinical features and results of Tier 1 testing
<ul style="list-style-type: none"> Chromosome microarray studies Fragile X Complete metabolic panel, CBC, CRP Amino acid level Lactate and pyruvate levels Carnitine, plasma total and free Catecholamine (G17 level) Plasma and urine amino acids Urine organic acids Plasma acylcarnitines Thyroid function tests 	<ul style="list-style-type: none"> Mitochondrial enzyme and/or DNA testing Rett syndrome DNA testing Appraisal Panel (CCK25) gene testing PIEN mutation analysis NIGNS3, NIGNS4X, SHANK3, SNRPN gene testing lysosomal enzyme testing Neuroscience disease testing (NIDA) CSF studies for lactate and pyruvate, amino acids and neurotransmitters Brain MRI

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Reference

December 1, 2010, Vol 304, No. 21 >

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Preliminary Communication | December 1, 2010

Mitochondrial Dysfunction in Autism FREE

Cecilia Giulivi, PhD, Yi-Fan Zhang, BS, Alicja Omanska-Klusek, MS, Catherine Ross-Inta, BS, Sarah Wong, BS, Inva Hertz-Picciotto, PhD, Flora Tassone, PhD, Isaac N. Pessiah, PhD

[*] Author Affiliations

JAMA. 2010;304(21):2389-2396. doi:10.1001/jama.2010.1796

Text Size: A A A

<http://jama.jamanetwork.com/article.aspx?articleid=186999>

Metabolic / Mitochondrial Disease

- The Giulivi study
 - Suggests a stronger link between ASD and Mitochondrial disease
- Need for screening for Mitochondrial dysfunction
 - Important for ER staff in times of illness and stress
 - Important for selecting anesthesia medications
 - Aggressive metabolic management improves quality of life
- SPECIALIST MUST HAVE EXPERIENCE DIAGNOSING & TREATING MITOCHONDRIAL DISEASE IN ASD

For more information:
www.mindinstitute.ucdavis.edu
www.my.clevelandclinic.org
www.mitoaction.org

Reference

Cerebral folate receptor autoantibodies in autism spectrum disorder

Open

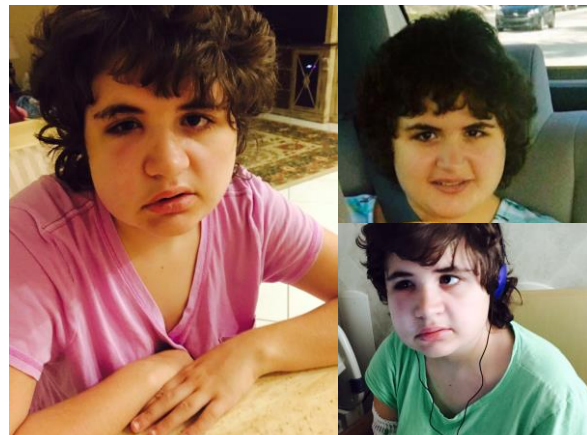
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<http://www.nature.com/mp/journal/v18/n3/full/mp2011175a.html>



Dental Problems as a Source of Pain

- Atypical presentations are common
- Change in chewing habits
- Self injury involving head and face
- Vital to obtain X-rays every 6-12 months
- Desensitize by frequent dental visits and oral hygiene
- Special needs dentist helpful in identifying problems and accessing appropriate anesthesia for thorough evaluation



Pressing Healthcare Needs

- "Development of standards of care for diagnostic recognition & treatment of medical problems in persons with ASD"
 - Margaret Bauman MD
 - Founder of the Ladders Program now The Lurie Family Autism Center
- "To not treat the comorbid medical conditions in persons with autism is medical neglect"
 - Vicki Martin RN
- High caliber clinical databases and TREATMENT based Centers of Excellence are needed in every state



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