#### 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.ccfcme.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



Recommendations for Evaluation and Treatment of Common Gastrointestinal Problems in Children With ASDs Timothy Buie, George J. Fuchs III, Glenn T. Furuta, Koorosh Kooros, Joseph Levy, Jeffery D. Lewis, Barry K. Wershil and Harland Winter *Pediatrics* 2010;125;S19 DOI: 10.1542/peds.2009-1878D

The online version of this article, along with updated information and services, is located on the World Wide Web at: /content/125/Supplement 1/S19.full.html

#### PEDIATRICS

OFFICIAL JOURNAL OF THE AMERICAN ACADEMY OF PEDIATRIC

Evaluation, Diagnosis, and Treatment of Gastrointestinal Disorders in Individuals With ASDs: A Consensus Report

Timothy Buie, Daniel B. Campbell, George J. Fuchs III, Glenn T. Furuta, Joseph Levy, Judy VandeWater, Agnes H. Whitaker, Dan Atkins, Margaret L. Bauman, Arthur L. Beaudet, Edward G. Carr, Michael D. Gershon, Susan L. Hyman, Pipop Jirapinyo, Harumi Jyonouchi, Koorosh Kooros, Rafail Kushak, Pat Levitt, Susan E. Levy, Jeffery D. Lewis, Katherine F. Murray, Marvin R. Natowicz, Aderbal Sabra, Barry K. Wershil, Sharon C. Weston, Lonnie Zeltzer and Harland Winter *Pediatrics* 2010;125;S1 DOI: 10.1542/peds.2009-1878C

The online version of this article, along with updated information and services, is located on the World Wide Web at: /content/125/Supplement 1/S1.full.html

### **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 disproportionally higher than in the general population

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

Gastrointestinal symptoms in ASD: a meta-analysis
 <u>www.pediatrics.aappublications.org</u>

## **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

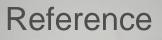


Information taken from: Neurological Comorbidities in ASD Margaret Bauman, M.D. www.ccfcme.org/autism15

# 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





#### frontiers in **PUBLIC HEALTH**

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

Richard E. Frye<sup>1</sup>\*, Daniel Rossignol<sup>2</sup>, Manuel F. Casanova<sup>3</sup>, Gregory L. Brown<sup>4</sup>, Victoria Martin<sup>4</sup>, Stephen Edelson<sup>5</sup>, Robert Coben<sup>6</sup>, Jeffrey Lewine<sup>7</sup>, John C. Slattery<sup>1</sup>, Chrystal Lau<sup>1</sup>, Paul Hardy<sup>8</sup>, S. Hossein Fatemi<sup>9</sup>, Timothy D. Folsom<sup>9</sup>, Derrick MacFabe<sup>10</sup> and James B. Adams<sup>11</sup>

- 1 Arkansas Children's Hospital Research Institute, Little Rock, AR, USA
- <sup>2</sup> Rossignol Medical Center, Irvine, CA, USA
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- <sup>4</sup> Autism Recovery and Comprehensive Health Medical Center, Franklin, WI, USA
- <sup>5</sup> Autism Research Institute, San Diego, CA, USA
- <sup>6</sup> New York University Brain Research Laboratory, New York, NY, USA
- <sup>7</sup> MIND Research Network, University of New Mexico, Albuquerque, NM, USA
- <sup>8</sup> Hardy Healthcare Associates, Hingham, MA, USA
- <sup>9</sup> University of Minnesota Medical School, Minneapolis, MN, USA
- <sup>10</sup> University of Western Ontario, London, ON, Canada
- " Arizona State University, Tempe, AZ, USA

<u>http://www.amazingbrains.com/wp-</u> <u>content/uploads/2014/05/article\_file\_4seizures-in-ASD.pdf</u>

## 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:
  - Otis 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**

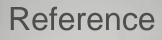
- PANDAS (Pediatric Autoimmune Neuropsychiatric Disorder Associated with strep)
- PANS (Pediatric Acute Neuropsychiatric Syndrome)
- Both are defined by 5 criteria:
  - Abrupt, significant onset of OCD and/or Tics
  - Includes other neuropsychiatric symptoms
  - Prepubertal onset
  - Association with 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: <u>www.nimh.nih.gov</u>





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**Consensus Statement** 

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

Kiki Chang, MD<sup>1,\*</sup> Jennifer Frankovich, MD<sup>2,\*</sup> Michael Cooperstock, MD, MPH<sup>3</sup>, Madeleine Cunningham, PhD<sup>4</sup>, M. Elizabeth Latimer, MD<sup>5</sup>, Tanya K. Murphy, MD<sup>6</sup>, Mark Pasternack, MD<sup>7</sup>, Margo Thienemann, MD<sup>8</sup>, Kyle Williams, MD<sup>9</sup>, Jolan Walter, MD<sup>10</sup>, and Susan E. Swedo, MD<sup>11</sup>; 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.ccfcme.org/autism15

### Bridging the Gap Between ASD and Mitochondrial disease By Fran Kendall, M.D.

### http://www.mitoaction.org/files/AutismOne2011.pdf

Table 1           Possible Symptoms of Mitochondrial Disease						
NERVES Absent reflexes Weakness (may be intermittent)	<ul> <li>Fainting</li> <li>Dysautonomia (e.g., temperature instability)</li> </ul>	•	Neuropathic pain			
MUSCLES VWeakness Cramping	<ul><li>Hypotonia</li><li>Muscle pain</li></ul>					
GASTROINTESTINAL Gastrointestinal problems Irritable bowel syndrome Dysmotility Pseudo-obstruction	<ul><li>Gastroesophageal reflux</li><li>Diarrhea or constipation</li></ul>					
<ul> <li>Renal tubular acidosis or wasting</li> </ul>						
<ul><li>HEART</li><li>Cardiomyopathy</li></ul>	Cardiac conduction defects (here	art blocks)				
LIVER Liver failure	<ul> <li>Hypoglycemia (low blood sugar</li> </ul>	Hypoglycemia (low blood sugar)				
EARS & EYES Visual loss and blindness Ptosis Ophthalmoplegia	<ul><li>Optic atrophy</li><li>Hearing loss and deafness</li></ul>	:	Acquired strabismus Refinitis pigmentosa			
PANCREAS & OTHER GLANDS     Diabetes and exocrine pancreatic for     Parathyroid failure (low calcium)	ailure (inability to make digestive enzymes)					
SYSTEMIC     Failure to gain weight     Fatigue	<ul> <li>Unexplained vomiting</li> <li>Short stature</li> </ul>		Respiratory problems			

### Bridging the Gap Between ASD and Mitochondrial disease By Fran Kendall, M.D.

### http://www.mitoaction.org/files/AutismOne2011.pdf

	Genetic Testing for Autis	Table 2	Disorder (ASD) Patients	
TIPD			10 10 10 10 10 10 10 10 10 10 10 10 10 1	
TIER 1 Basic workup recommended for all patients		TIER 2 Dependent on clinical features and results of Tier 1 testing		
-	Chromosome microarray studies		Mitochondrial enzyme and/or DNA testing	
-	Fragile X		Rett syndrome DNA testing	
-	Complete metabolic panel, CBC, CPK	-	Atypical Rett (CDKL5 gene testing)	
-	Ammonia level	-	PTEN mutational analysis	
-	Lactate and pyruvate levels		NLGN3, NLGN4X, SHANK3, SNRPN gene testing	
-	Carnitine, plasma total and free	-	Lysosomal enzyme testing	
-	Coenzyme Q10 level		Peroxisome disease testing (VLCFAs)	
-	Plasma and urine amino acids		CSF studies for lactate and pyruvate, amino acids	
-	Urine organic acids		and neurotransmitters	
-	Plasma acylcarnitines		Brain MRI	
-	Thyroid function tests			

## Reference

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

< Previous Article Next Article >

Preliminary Communication | December 1, 2010

### Mitochondrial Dysfunction in Autism

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

#### [+] Author Affiliations

JAMA. 2010;304(21):2389-2396. doi:10.1001/jama.2010.1706.

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### 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: <u>www.mindinstitute.ucdavis.edu</u> <u>www.my.clevelandclinic.org</u> <u>www.mitoaction.org</u>

## Reference

# Cerebral folate receptor autoantibodies in autism spectrum disorder

#### Open

#### R E Frye<sup>1</sup>, J M Sequeira<sup>2</sup>, E V Quadros<sup>2</sup>, S J James<sup>1</sup> and D A Rossignol<sup>3</sup>

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Received 4 August 2011; Revised 3 November 2011; Accepted 7 November 2011 Advance online publication 10 January 2012

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

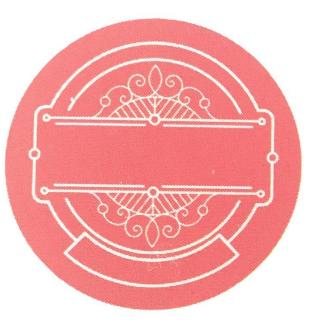


## **Dental Problems as a Source of Pain**

- 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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