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The Spectrum of Medical Child Abuse and What is Happening Across the Country?

Richard G. Boles, M.D.
Director, CNNH NeuroGenomics Program
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Medical Geneticist in Private Practice
Faculty Advisor, MitoAction
MitoAction Webinar
May 4, 2018

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Disclosure: Dr. Boles wears many hats

- Clinician treating patients
  - Primary interests in functional disease (autism, cyclic vomiting)
  - Past: Geneticist/pediatrician 20 years at CHLA/USC
  - Present: Director, CNNH NeuroGenomics Program (https://cnnh.org/cnnh-services/neurogenomics-program)
  - Present: In private practice in California (http://molecularmitomd.com)
- Chief Medical & Scientific Officer of NeuroNeeds LLC
  - Present: The company that produces SpectrumNeeds™ (https://neuroneeds.com)
- Medical Director for DNA Sequencing Companies
  - Past: 5 years at Courtagen Life Sciences; 6 months at Lineagen
  - Present: Loose affiliations with some companies
- Expert witness in legal cases
  - Present: Medical child abuse, child neglect and custody cases (drboles@molecularmito.com)
  - Vaccine Court, malpractice cases
- Researcher with prior NIH and foundation funding
  - Past: USC faculty for 20 years
  - Present: Study sequence variation that predispose towards neurodevelopmental and functional disorders

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Case Report 1
Alejandro – early 1990s

1. 3-year-old boy with mild developmental delay, hypotonia, and muscle weakness
2. Probable mitochondrial disease diagnosis based on phenotype, biochemistry, apparent maternal inheritance, and apparent clinical improvement with cofactor therapy
    - 7 siblings and mother all with some degree of developmental delay
3. Mother was disowned by her own mother and she and her kids were cut off from the extended family.
4. Child developed episodes of hypernatremia (high blood sodium/salt), alkalosis, and vomiting blood.
    - 3 episodes were life-threatening; sent to ICU
    - Several milder episodes that mostly led to bloody diarrhea
    - Extended family rallied around and supported the mother
5. No apparent medical cause was identified.
6. A much-younger Dr. Boles was the attending physician.

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6. A much-younger Dr. Boles was the attending physician.
7. Blood testing via indwelling catheter every 4 hours revealed normal findings just prior to a life-threatening event, in which case the blood sodium and bicarbonate levels were extremely elevated.
8. Video-EEG was ordered. No further events occurred.
9. The monitor was turned off, but the camera was left on. Video

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7. Blood testing via indwelling catheter every 4 hours revealed normal findings just prior to a life-threatening event, in which case the blood sodium and bicarbonate levels were extremely elevated.
8. Video-EEG was ordered. No further events occurred.
9. The monitor was turned off, but the camera was left on. Video showed mom mixing a substance and injecting into the G-tube through a very large syringe.
10. Security found empty Alka-Seltzer tins in mom's purse, and the large syringe.
    - Alka-Seltzer is sodium bicarbonate and aspirin. Aspirin can cause stomach ulceration with bleeding.
11. Mother charged with attempted murder, pleaded guilty, and sentenced to hard time.
12. Child stable several years later.
    - Still with same degree of developmental delay and hypotonia.

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Case Report 1
Alejandro – early 1990s

1. Some people really do abuse their children.
2. A pediatrician is charged with both protecting his patient (the child) and in protecting the family.
    - When these are in conflict, the child takes precedence.
3. Most cases of medical child abuse (MCA) occur in children who have a real underlying medical condition.
4. Actually having mitochondrial disease, or any other condition, even if one can prove it, does NOT negate the possibility of MCA.
5. The key is to whether mitochondrial disease, or other diagnosis, can account for the issues that are of concern.

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Case Report 2
Bailey – early 2010s

1. A previously-normal, intelligent, early-adolescent girl suffered from multiple functional symptoms that started following an apparent stomach flu at age 10 years:
    - Severe dysmotility, which has required long-term TPN
    - Chronic fatigue syndrome
    - Chronic pain in multiple locations
    - Unexplained tachycardia
2. Probable mitochondrial disease diagnosis based on phenotype, biochemistry, apparent maternal inheritance, apparent clinical improvement with cofactor therapy, and a variant of uncertain significance in the mtDNA thought to be disease related.
3. Additional features developed:
    - Anemia requiring multiple transfusions
    - Severe dysautonomic reactions often mimicking septic shock with negative cultures (shock syndrome)
4. Local physicians could not identify an apparent medical cause for neither the severe anemia nor the life-threatening episodes of shock.

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4. Local physicians could not identify an apparent medical cause for neither the severe anemia nor the life-threatening episodes of shock.
5. Suspected MCA was reported.
    - The family traveled to another state for expert evaluation.
    - Suspected MCA was reported again.
    - The patient, then in her mid-teens, was held against her will in the hospital for months.
    - The mitochondrial cocktail was discontinued.
    - The parents were not allowed to contact her.
    - The hospital and physicians did not attempt to communicate with her previous medical providers.

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    - The mitochondrial cocktail was discontinued.
    - The parents were not allowed to contact her.
    - The hospital and physicians did not attempt to communicate with her previous medical providers.
6. The patient's condition deteriorated throughout the time of separation.
    - Under increasing legal pressure, the family was allowed to transfer the patient to the care of a friendly local physician.
    - The mitochondrial cocktail was reestablished.
    - The patient improved dramatically.
7. Exome sequencing revealed mutations in:
    - A mtDNA gene, likely accounting for mitochondrial dysfunction (previously identified)
    - A connective tissue/Ehlers-Danlos gene, likely accounting for dysautonomia
    - A Blackfan-Diamond gene, likely accounting for severe anemia
8. Several years later:
    - Bailey is clinically stable.
    - Has a few hospitalizations each year for blood transfusions and line issues.
    - Attending university full-time
    - All legal action against the family was dropped.

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Case Reports 1 and 2 Analysis

1. While very different outcomes, both cases had similar beginnings:
   - Non-specific clinical findings
   - A clinical diagnosis of mitochondrial disease that was not proven, and was challenged by some physicians.
   - Later development of severe life-threatening intermittent manifestations
   - An inability of the physicians to medically account for these later developments
     - Dysautonomic reactions can occur, but are not usually this severe.
     - Anemia can occur, but is not usually this severe.

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2. How are the cases different? What was missed in the second case?

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2. How are the cases different? What was missed in the second case?
   - There was no communication with previous physicians.
   - There was no attempt to order additional testing.
   - Even in the face of continued deterioration, the hospital continued with its assessment and plan.

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3. Were the rights of the patient and/of family violated in either case? If so how? What would you suggest should have been done instead?
   - Case 1
   - Case 2

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Case Report 3
Caitlyn – early 2010s

1. 6-year-old girl with:
   - Intestinal pseudoobstruction
   - Chronic pain syndrome including somatic and visceral hypersensitivity
   - Generalized dysautonomia, including unexplained tachycardia
   - ADHD
2. Probable mitochondrial disease diagnosis based on phenotype, apparent maternal inheritance, muscle biopsy, and apparent clinical improvement with cofactor therapy
3. Caitlyn presented to the ER multiple times and her care continued to escalate:
   - Feeding tolerance worsened so that she was on tube feedings, TPN, and frequent additional IV fluids
   - Multiple hospitalizations for fever and/or tachycardia, r/o line sepsis
   - BiPAP and supplemental oxygen
   - IVIG
   - Narcotiics
4. Presented to another facility. No apparent medical cause was identified to justify the escalation in care management.

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   - BiPAP and supplemental oxygen
   - IVIG
   - Narcotiics
4. Presented to another facility. No apparent medical cause was identified to justify the escalation in care management.
5. Suspected MCA was reported.
   - The child was held in the hospital against her mother's will for months.
   - Limited and supervised visitation was instituted.
   - The hospital and physicians had limited communication with her previous providers.
   - She was successfully weaned off of several aspects of her pervious medical care, including narcotics, BiPAP, supplemental oxygen, IV fluids, TPN, tube feedings, and IVIG.
   - The diet was expanded.
   - The mitochondrial cocktail was discontinued.
   - She remained in good condition throughout the admission and weaning process.

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Case Report 3 Analysis

1. Again this case has similar beginnings to the previous cases:
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2. What are the medical and legal teams not considering herein?

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2. What are the medical and legal teams not considering herein?
   - The mother never misrepresented herself, although her assessments regarding the child's discomforts were likely off.
   - Each and every escalation was performed by medical professionals.
   - All of the providers were aware of what other providers had done, or should have been.
3. Who is responsible for the unnecessary escalation of care in the absence of fraud?

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2. What are the medical and legal teams not considering herein?
   - The mother never misrepresented herself, although her assessments regarding the child's discomforts were likely way off.
   - Each and every escalation was performed by medical professionals.
   - All of the providers were aware of what other providers had done, or should have been.
3. Who is responsible for the unnecessary escalation of care in the absence of fraud?
4. Following my testimony, the child was returned to the family under the legal responsibility of the maternal grandparents regarding medical care, but with no further restrictions on mom's involvement.
   - Child is clinically stable and requires no ER visits.
   - Still has functional symptoms related to some residual pain and fatigue.
   - No care is now provided other than medications for ADHD, dysmotility and neuropathic pain.
   - Regular diet without supplementation.

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5. What can we learn regarding what happened in this case?

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Case Report 3 Analysis

- Can a good parent be contributing to harm even if they always had the best intentions and never lied?

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Analysis of Cases 1-3 and About One Hundred Others

- Non-specific clinical findings
  - GI failure on tube feedings or TPN
  - Multiple hospitalizations with fever and tachycardia, rule/out sepsis
  - Organ failure, especially bone marrow with anemia and infections
- A controversial diagnosis in the absence of proof
- Severe, even life-threatening, intermittent manifestations
  - Protean
- An inability of the physicians to medically account for the issues of concern
  - In part due to extreme biological and clinical complexity
  - In part due to a failure of the Western Medical Model
- Mental health issues, including anxiety and/or depression, in the child and other family members.
  - Especially anxiety and/or depression in the mother
- A breakdown in communication, respect, and/or support between the family and one or more care providers.
  - In most of these cases, there are MULTIPLE care providers given the number of specialists referred and the rotation of services.
- Presentation to another medical facility, for a “second opinion”, procedure, or emergency.

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What Can You Do? Pre-Allegation

- A team leader – one person who runs the show
  - Assemble a team that works with the leader and with each other.
  - Work collaboratively with the leader and team, but let them take the heat.
  - Do not doing anything important without the input of the team leader.
  - Have the important information with you at all times.

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- Be very careful regarding presenting at other facilities
  - Clear it first with the team leader, or at least with a team member.
  - By very careful while traveling.

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- Insist on DNA testing, at least including exome and mtDNA
- Avoid breakdowns in communication and respect.
  - Always treat the medical team with respect, especially if they do not deserve it.
  - You do not have to agree; you do not have to comply, but you must keep an open line of communication.
  - Let your team leader take the heat.

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  - Always treat the medical team with respect, especially if they do not deserve it.
  - You do not have to agree; you do not have to comply, but you must keep an open line of communication.
  - Let your team leader take the heat.
- If you are affected, get treatment
  - Especially regarding anxiety and/or depression in the mother
  - You cannot adequately advocate for your child if you are not healthy yourself.

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What Can You Do?
Post-Allegation

- Don’t panic!

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What Can You Do?
Post-Allegation

- Don’t panic!
- What happens when they call in the social worker?
  - Tell the truth. Explain why you are anxious. Explain what you want.
  - Be in problem-solving mode.

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What Can You Do?
Post-Allegation

- Don’t panic!
- What happens when they call in the social worker?
  - Tell the truth. Explain why you are anxious. Explain what you want.
  - Be in problem-solving mode.
- Put yourself in their shoes
  - The child is very sick and they cannot seem to help.
  - If they have concerns that you may be part of the problem, they are legally and morally required to act.
  - They may be ignorant, but they are not evil.

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  - They may be ignorant, but they are not evil.
- Involve your medical team “early and often”.
- If a report is filed, get legal counsel.
  - Insist on an expert witness if there is legal action.

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Beyond the Metabolic Pathways

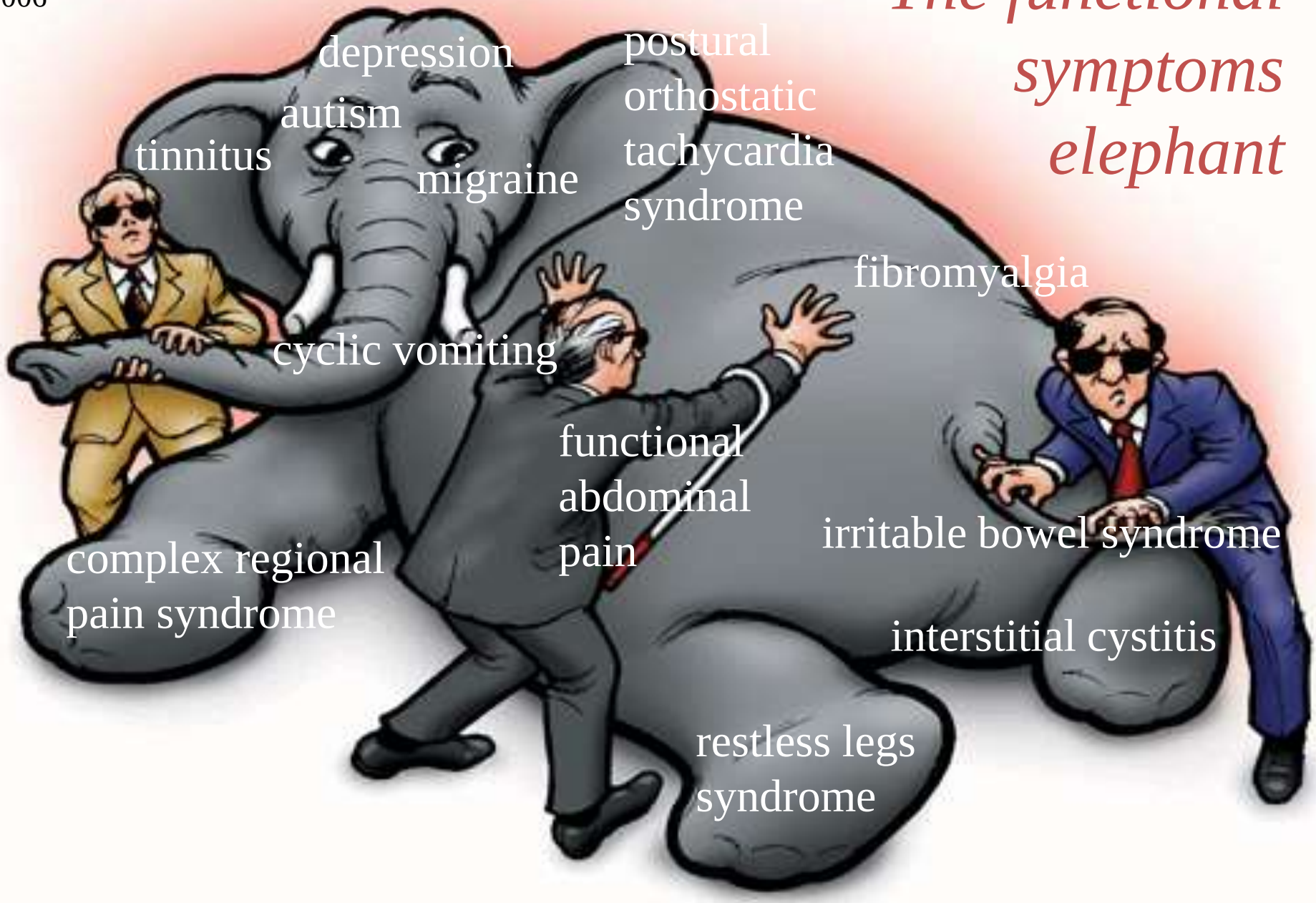
- There are hundreds of known causes of mitochondrial disease
- There are perhaps more causes of Secondary mitochondrial dysfunction
- In most patients the underlying gene is not obvious even by an expert.
- The complexity lends itself to massive parallel sequencing = “NextGen sequencing”.

Not on this slide:

- Transcriptional elements
- Translational elements
- Chaperones
- Glycosylation
- Assembly factors
- Other post-translational elements
- Mitochondrial Import
- Cofactor metabolism
- Antioxidant pathways
- Many others
- Causes of secondary mitochondrial dysfunction
  - Ion channels
  - Peroxisomal biogenesis factors
- Many others
- Phenocopies

Gardner
Boles
2006

The functional symptoms elephant



The elephant is lying down due to chronic fatigue

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20 "Functional" Disorders:

- Attention deficit hyperactivity disorder
- Anxiety disorder
- Autistic spectrum disorders
- Chronic fatigue syndrome
- Complex regional pain syndrome
- Cyclic vomiting syndrome
- Depression (MDD)
- Fibromyalgia
- Functional abdominal pain
- Interstitial cystitis
- Insomnia (chronic, severe)
- Irritable bowel syndrome
- Migraine
- Panic disorder
- Post-traumatic stress disorder
- Postural orthostatic tachycardia syndrome
- Restless legs syndrome
- Temporomandibular disorder
- Tinnitus
- Vulvovaginitis syndrome

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What Is Functional Disease?

A poem by a 14-year-old patient

I never know when its going to come back
This fatigue is an internal attack
It so easily cripples me
Only no one can see

Its so hard when you easily tire
And everyone around you thinks your lazy and a liar
They cant see so they don't know
I know in my heart its real though

Its a relief to get the answer and know you're not crazy
You can finally prove you're not just lazy
Its still not easy and never will be
But maybe some day the world will see

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Don’t Panic!