Congenital Central Hypoventilation Syndrome (CCHS)

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http://cchsnetwork.org

Disclosures

- Research support Eli Lilly, ASCO, AACR
- Advisory board STRATA Oncology

Madi's journey

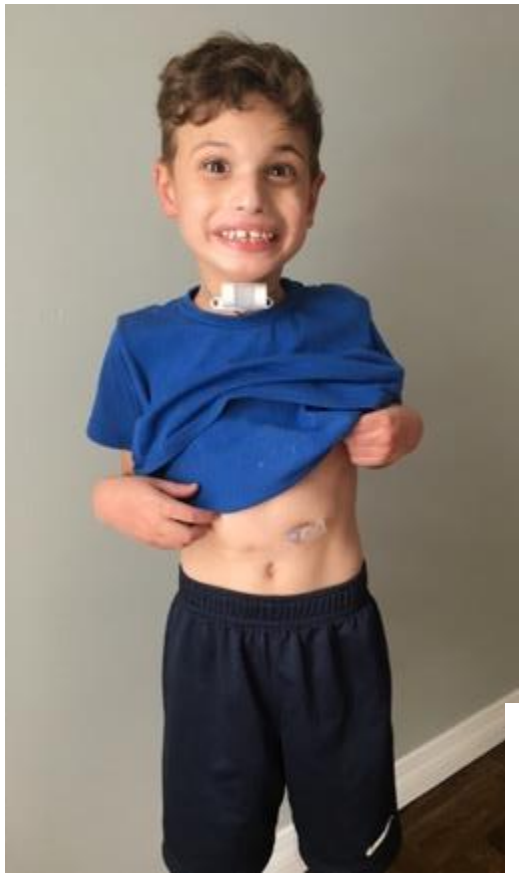
- Born Jan 19, 2011
- 6 months in the NICU – trach, O2 sats, transdermal CO2 monitor, NG feeds
  - Diagnosed with CCHS Feb 11th
- PT/OT/Swallow therapy

Madi's journey

- 1st 2 years vent dependent
  - Repeat hospitalizations
  - Multiple "blue spells" → seizures
- Jan 1st 2013 – syncopal episode
  - Cardiac Pacemaker Jan 4th
- Speech, reading delay
- GH deficiency

What is CCHS?

- ***Rare congenital*** disorder that affects multiple systems
  - Cardiac
  - Gastrointestinal
  - Pulmonary
  - Nervous system
  - Endocrine
- ICD-9 CM: 327.25
- ICD-10 CM: G47.35
- Ondine's curse, central alveolar hypoventilation syndrome



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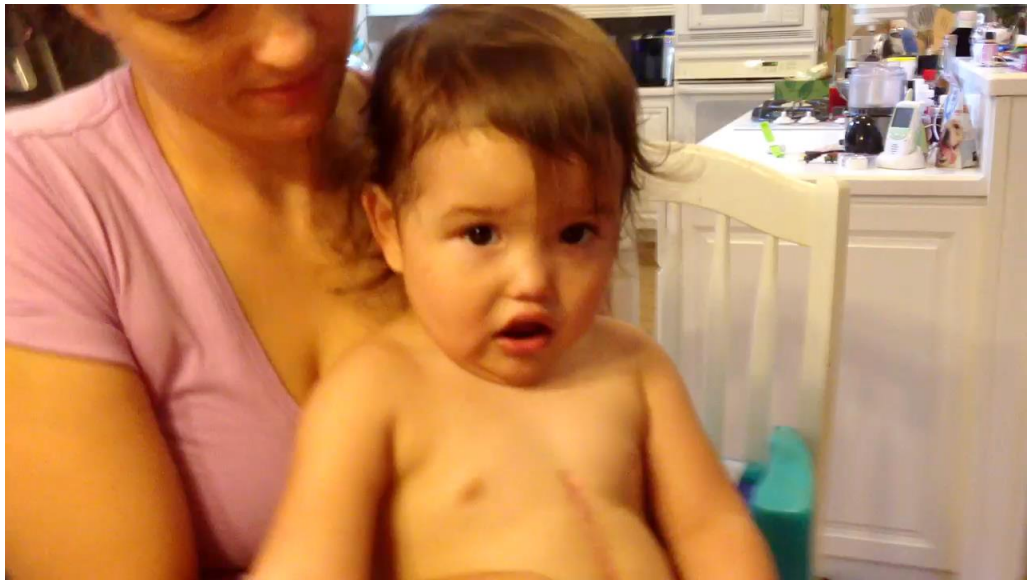
What is CCHS?

- Diagnostic testing
  - PHOX2B testing
  - Rectal biopsy
  - 24hr holter
  - Neuroblastoma screening
- Physical findings
  - Lack of response to hypoxia or hypercapnia
  - Cardiac pauses
  - Hirschsprung's disease
  - General dysautonomia
  - Cognitive impairment
  - Developmental delays



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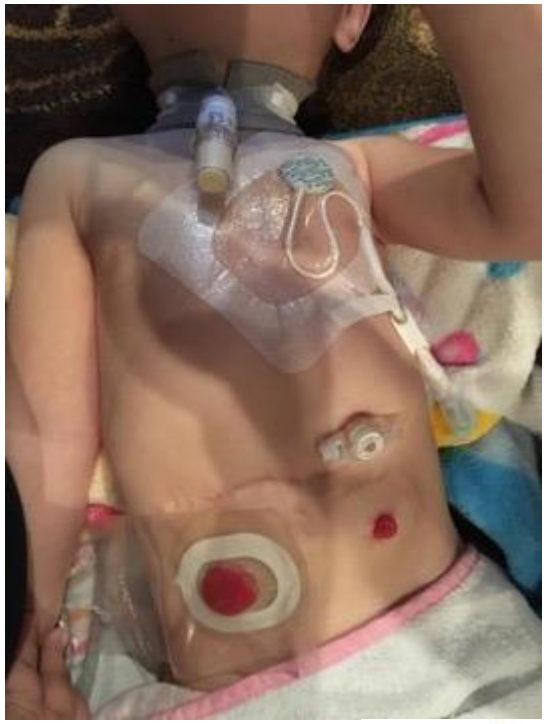
What is CCHS?

- Severity
  - Heterogeneous
  - Life long medical assistance
    - Professionals,
    - Parents, caregivers, spouses
- Progression
  - Unknown
  - NORD Registry
- Treatment
  - No cure
  - Supportive care
- Response to treatment – N/A

Systems affected

- Pulmonary (vent dependence, repeated pneumonias, hospitalization)
- Cardiac pauses
- GI (hirschsprung's, reflux, feeding difficulties)
- Increased risk of neuroblastoma
- Eye problems
- Neurodevelopmental delays
- Physical delays
- Speech delays



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Patient/family experiences

"Our kids tend not to speak up about this most important reality."

"I am afraid to tell anyone about my condition because it will be used against me and my co-workers will look at me/treat me differently"

"She is living on her own and no one knows about her condition or what to do in case of an "event" - ***could be catastrophic***....I live in fear of *that* phone call."

"He is really struggling with depression... He is now back home, trying twice to be on his own but financially not able to do so."

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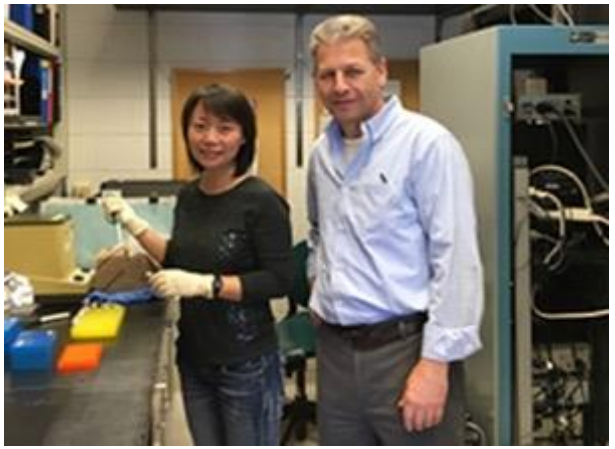
Research

- NORD Registry
- CCHS Foundation
  - Douglas Bayliss
  - Isabella Ceccherini
  - Diego Fornasari
  - Namasivayam Ambalavanan



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SSA criteria

- **103.02 Child respiratory disorders**
  - The presence of a tracheostomy.
  - Upon the attainment of age 3, documented need for mechanical ventilation via a tracheostomy for ***at least 4 hours per day*** and for at least 90 consecutive days.

- **103.14 Respiratory failure**
  - Chronic respiratory disorder except CF
    - Requiring invasive mechanical ventilation, noninvasive ventilation with BiPAP, or a combination of both treatments,
    - Continuous period of at least 48 hours, or for a continuous period of at least 72 hours if postoperatively, twice within a 12-month period and at least 30 days apart

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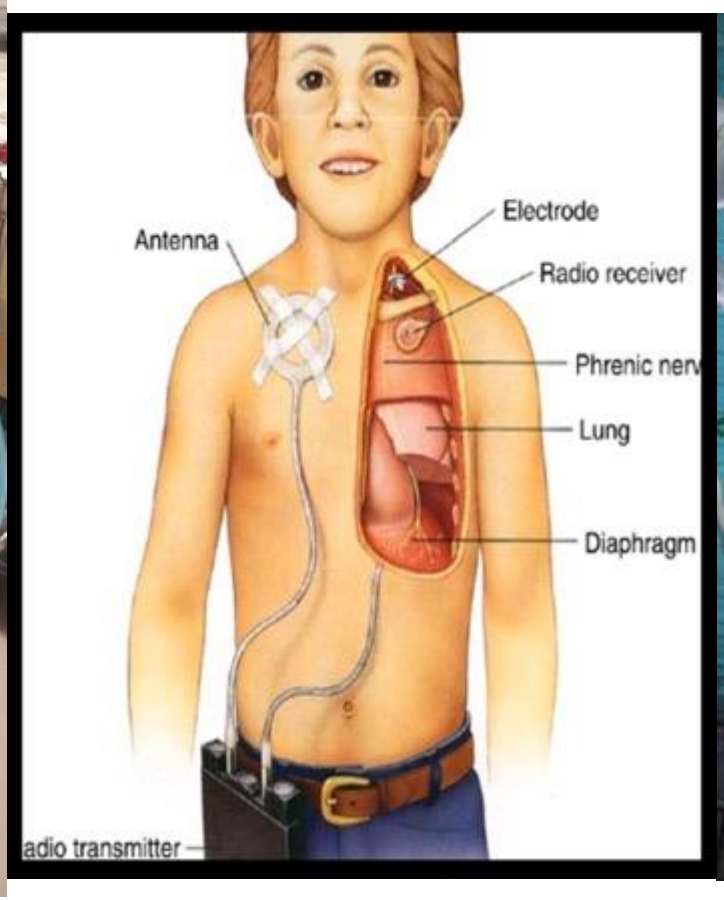
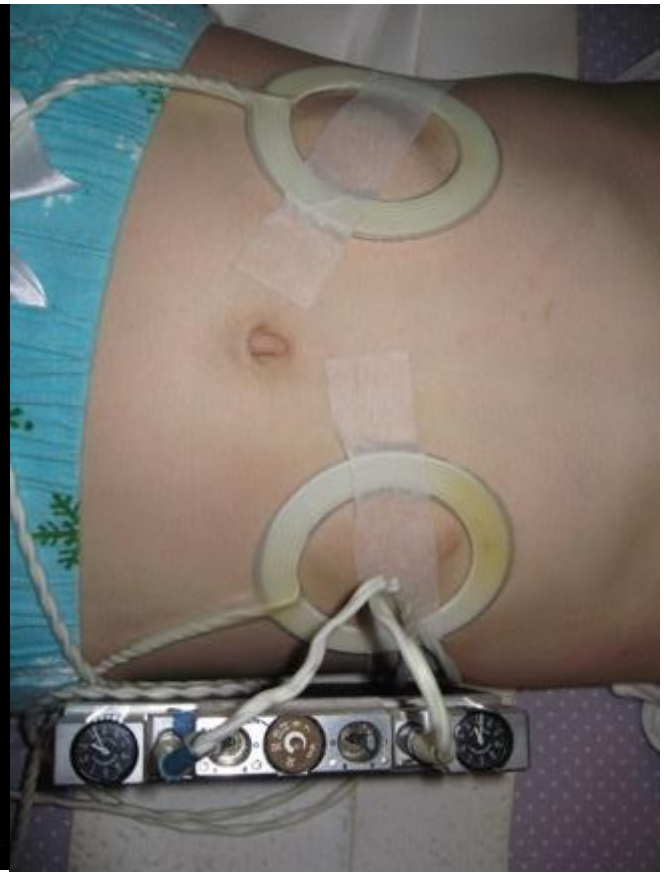
Respiratory assistance





Other SSA criteria

- **109.00 Endocrine Disorders**– hyperinsulinemia , and GH deficiencies noted in the population
- **105.07 Short bowel syndrome** – Hirschsprung's
  - 105.10 Need for supplemental daily enteral feeding via a gastrostomy
  - Some kids require total parenteral nutrition via central line
- **111.09 Communication impairment**, associated with documented neurological disorder and one of the following:
  - Documented speech deficit that significantly affects (see 111.00K1) the clarity and content of the speech.
    - Tracheostomy, dysautonomia
    - Apraxia

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Other SSA criteria

- *Intellectual disorder (112.05).*
- *Anxiety and obsessive-compulsive disorders (112.06).*
- *Autism spectrum disorder (112.10).*
- *Neurodevelopmental disorders (112.11).*
- *Developmental disorders in infants and toddlers (112.14).*

CCHS is a rare congenital multi-system disorder

- **<u>10.00 Congenital Disorders that Affect Multiple Body Systems</u>**
  - Severe medically determinable impairment(s) that does not meet a listing
  - May or may not have the residual functional capacity to engage in substantial gainful activity.
- *Other congenital disorders that affect multiple body systems.*
  - cause deviation from, or interruption of, the normal function of the body or can interfere with development
  - degree of deviation, interruption, or interference, as well as the resulting functional limitations and their progression, may vary widely from person to person and may affect different body systems.

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Summary

- CCHS is a RARE lifelong congenital multi-system condition that requires medical assistance/support
- Affects multiple systems resulting in multiple co-morbidities
- Addition to the list of compassionate allowances condition would benefit the CCHS patients and families



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Acknowledgements

- CCHS Network
- CCHS families and patients
- SSA disability forum organizers

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