



Neurology in DESSH Syndrome

SHEEL PATHAK, MD
DEPARTMENT OF NEUROLOGY | DIVISION OF PEDIATRIC AND DEVELOPMENTAL NEUROLOGY
WASHINGTON UNIVERSITY SCHOOL OF MEDICINE

SECOND ANNUAL FAMILY CONFERENCE

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Disclosure Statement

Dr. Pathak is an Assistant Professor of Neurology and Pediatrics, Employed by Washington University

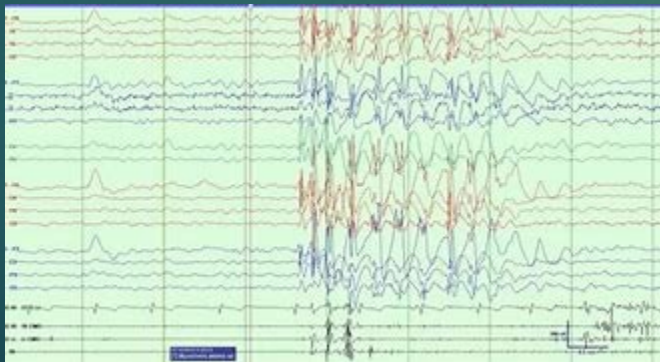
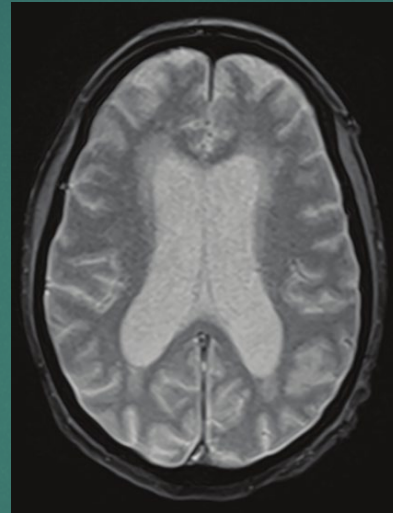
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Financial — Dr. Pathak has no financial interest, arrangement, or affiliation which would constitute a conflict of interest

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Neurologic Manifestations of DeSanto-Shinawi Syndrome



Developmental Diagnoses – Brief Descriptions

- ▶ Global Developmental Delay (GDD)
 - ▶ Young Child (<5) with delays in multiple areas (fine motor, gross motor, language, social/adaptive)
 - ▶ Periodically tracking development is important
- ▶ Intellectual Disability (ID)
 - ▶ Deficits in:
 - ▶ Intellectual functions (problem-solving, planning, abstract thinking, academics)
 - AND
 - ▶ Adaptive functions (ability to function at level of independence expected for age)
 - ▶ Can be mild, moderate, severe, or profound
- ▶ **Development typically goes forward. Regression is not normal!**

Management of Developmental Disorders

▶ Medications

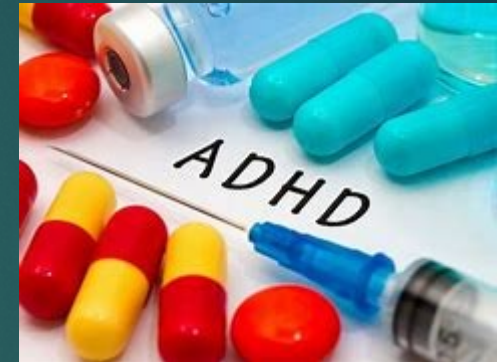
- ▶ Can treat ADHD or certain types of harmful behaviors with medication

▶ Optimize further development

- ▶ Ensure vision and hearing are good
- ▶ Sleep!
- ▶ Nutrition – healthy, balanced diet, multivitamin if child is extremely picky

▶ Therapies

- ▶ Physical therapy
- ▶ Occupational therapy
- ▶ Speech therapy
- ▶ Behavioral therapy
- ▶ Educational supports – Individualized Education Plan, “504 Plan”



Seizures/Epilepsy

- ▶ Seizure: Event caused by unhealthy electrical activity in the brain
- ▶ Epilepsy: predisposition to have seizures
 - ▶ 2 or more “unprovoked” seizures
 - ▶ 1 seizure + high risk of more
 - ▶ The description of the episodes are the most important!
 - ▶ EEG is helpful but NOT most important
 - ▶ Seizures are treatable (but not all require treatment)



Role of EEG (Electroencephalogram)

- ▶ EEG – a test that measures electrical activity in the brain at the time of the study

EEG CAN...	EEG DOESN'T
<ul style="list-style-type: none">- Identify a risk of seizure- Tell where seizures come from- Identify specific patterns- Help classify events if they happen <u>DURING</u> the test	<ul style="list-style-type: none">- Predict the next seizure- Tell if a seizure happened in the past

* Routine EEG can only address the surface of the brain!



When Should an EEG Be Performed?

- ▶ When there are **new** abnormal spells
- ▶ Before medication changes
- ▶ To capture a spell
- ▶ When considering brain surgery to treat epilepsy

Epilepsy in DESSH Syndrome

- ▶ Not everyone has epilepsy
- ▶ **Common:** Treatable with medication
- ▶ **Uncommon:** “Refractory” epilepsy
- ▶ In general, epilepsy should present in childhood and by adolescence at the latest.
- ▶ This is still a rare syndrome so we do not have good statistics yet.

Neurologic Management in DESSH Syndrome

- ▶ Neurologist:
 - ▶ Monitor Development
 - ▶ Medication Management (**seizures**, behavior)
 - ▶ Connects patients to therapies
- ▶ “Refractory” Epilepsy may need:
 - ▶ Special Diet (Ketogenic Diet, Modified Atkins Diet)
 - ▶ Surgery, Implants (VNS, RNS, DBS)
- ▶ Goal: ~~SEIZURES~~
- ▶ Assemble Your Team!
 - ▶ Therapists
 - ▶ Medical Team (Communicative, Responsive, Advocates)
 - ▶ School, Community
 - ▶ Support Groups



Questions?

- ▶ Judy Weisenberg, MD (314) 454.6120
 - ▶ Epilepsy Specialist
- ▶ Sheel Pathak, MD (314) 454.6363
 - ▶ General Pediatric Neurologist

