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Expanding the Phenotype of HNRNPU-related Disorders to Include Brief, Resolved, Unexplained Events (BRUE)

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Program: Genetics

Type: Case Report

Background: hnRNP-U deficiency is caused by pathogenic variants in HNRNPU, which encodes the heterogeneous nuclear ribonucleoprotein U (HNRNPU), a highly conserved protein responsible for assisting spliceosomes in mediating transcription and alternative splicing activity. HnRNPs are responsible for the regulation of translation at the presynaptic sites as well as the transportation of stabilized mRNAs along the axonal cytoskeleton.

Case: Here, we report a 2-year-old-male with a HNRNPU variant with a new presentation of apparent recurrent apneic spells with an underlying epileptic origin. These were described as apnea followed by desaturation and tachycardia in the 180's-200 range prior to resolution of symptoms. He also had autistic behaviors, hypotonia, global developmental delay, heart defects, and unique facial features. The anesthetist professional parents describe multiple BRUE.

At 26 months, he presented to the hospital with hypotonia and unique facial features, global developmental delay, autistic behaviors, dyspraxia with cognitive disability and a change in mental status. On physical exam, the proband had telecanthus, a broad nasal bridge, short palpebral fissures, mild nevus flammeus changes on his face, a single right palmar crease, and a modified single crease on the left. EKG showed a sinus rhythm with intermittent 1st degree AV block, blocked premature atrial contractions, left axis deviation, right bundle branch block, and an ejection fraction of 67%. Echocardiography re-identified an atrial septal defect. Brain MRI showed a T2/FLAIR hyperintense signal in the white matter of the parietal lobes, left greater than right. EEG identified generalized slowing indicative of mild nonspecific encephalopathy. The history of episodes was determined to be consistent with partial onset seizures with eye opening, deviation, and tachycardia with apnea and medical treatment ensued.

Genetic testing including microarray and an epilepsy panel identified no genomic dosage anomalies and a de novo nonsense mutation

(c.803+2T>C; p. unknown in HNRNPU), classified as pathogenic. Patient consent was obtained to use this case for educational purposes.

Conclusion: The study of hnRNP complexes has gained momentum in neurodegenerative and tumorigenesis disease research. hnRNPs have a key role in mediating transcription, alternative splicing, and translation activity. Recent research suggests 21 previously unreported probands; nearly doubling the recorded patient population. Probands in the literature to date have had variable presentation, but usually with hypotonia, global developmental delays, and seizures. This suggests the addition of HNRNPU to all seizure-related diagnostic panels. We would also recommend including the HNRNPU-related disorders in a differential diagnosis of BRUE and recurrent apneic episodes as any underlying clonic activity may be profoundly subtle. ■

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Presurgical Evaluation of the Dominant Hemisphere Function in an Adolescent Patient with Rasmussen's Encephalitis using Magnetoencephalography

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Program: Neurological Sciences

Type: Case Report

Background: Rasmussen's encephalitis (RE) is a devastating progressive inflammatory disease causing debilitating neurological deficits and intractable focal epilepsy. The therapeutic benefits of hemispherectomy in RE have been well established at this time; however, some patients do not achieve optimal seizure control following the surgery. The reasons for such suboptimal outcomes have included the presence of bilateral epileptogenic foci and involvement of the

dominant language hemisphere leading to concerns regarding surgical implications on preserved cognition, motor functions and language. There are currently no effective diagnostic tools to provide quantitative data regarding the potential deficits from the disconnection procedures in patients with RE.

Case: We present a case of a 15-year-old male with RE, right hemiparesis, recurrent status epilepticus and Epilepsia Partialis Continua who was considered for functional hemispherectomy of the dominant hemisphere. Due to his low baseline ambulatory function, and inability to cooperate with WADA test requirements, the non-invasive magnetoencephalography

(MEG) functional study was performed. This provided quantitative assessment of the lateralization of his language function and of his motor cortical organization to derive predictions for post-operative deficits. Using a passive listening auditory language paradigm, developed at our institution, the cortical responses to verbal stimuli were recorded within 350-450 ms in the right hemisphere and 600 ms in the left hemisphere, suggesting that language function was represented bilaterally. The source localization analysis confirmed that response in the right hemisphere was localized to the temporal-parietal area (angular gyrus), while response in the left hemisphere was localized to the posterior middle temporal gyrus.

*Names in bold type indicate presenting author.

These findings allowed us to appropriately counsel the patient's parents and advance his candidacy for surgical treatment. Following the hemispherectomy the patient's status epilepticus has resolved, and he has remained seizure free for 4 weeks. Post-operatively his hemiparesis and language function have improved. Patient consent was obtained to use this case for educational purposes.

Conclusion: To our knowledge, this is the first report on the use of MEG and a passive

listening paradigm in an adolescent patient undergoing presurgical evaluation for treatment of RE in a dominant hemisphere. The data obtained during this evaluation further broadens the utility of the MEG in providing valuable information regarding cortical reorganization of language function in progressive neurological conditions of an adolescent brain. The applications of MEG for mapping eloquent cortical functions may

be expanded to other chronic structural and autoinflammatory disorders of the brain. ■

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Non-inflammatory Bullae of the Dorsal Hand

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Type: Case Report

Background: Pseudoporphyria is a photodistributed disorder with clinical and histologic features similar to porphyria cutanea tarda. It is differentiated by having normal plasma, urine, and stool porphyrins. Common causes of pseudoporphyria include medications, sun exposure, and chronic renal failure. Young female patients are predominately affected.

Case: A 47-year-old male with acute myeloid leukemia status post allogeneic peripheral stem cell transplant (PSCT) developed a rash (scalp, upper neck, hand erythema worse with sun exposure) on day 16 post-transplant. Notable medications included voriconazole for fungal prophylaxis. Exam noted blanchable erythema over the neck and bilateral upper extremities with tense bullae over the dorsal left-hand 2nd and 5th digits. Punch biopsies for hematoxylin and eosin (H&E) and direct immunofluorescence (DIF) from the left-hand 2nd digit bullae and perilesional, respectively, revealed a subepidermal bullae with eosinophilic hyaline-like material on the roof, rete ridge festooning, and mild perivascular staining for IgG. This was consistent with porphyria or pseudoporphyria. Labs were obtained to differentiate the two, revealing normal serum and urine porphyrin levels. Given the constellation of findings, pathology, and laboratory, a diagnosis of pseudoporphyria secondary to voriconazole was made. Patient consent was obtained to use this case for educational purposes.

Conclusion: Pseudoporphyria develops most often secondary to non-steroidal anti-inflammatory drugs (NSAIDs). Voriconazole-



Figure 1. Panel A and C: Blanchable erythema on the neck and bilateral upper extremities. Scale in the postauricular regions. Panel B: Left dorsal hand with tense bullae overlying an erythematous base on the 2nd and 5th digits. Panel D: Demarcation over the right medial malleolus at the site of a previous sunburn.

induced cutaneous reactions occur in less than 10% of patients. Treatment options include discontinuation of causative agents, sunscreen application, and usage of sun-protective clothing. ■

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