

COMMUNICATIONS

Could it be Glut1?

Seizures that can't be seen on an EEG. Fasting and exercise intolerance. Fatigue. Leg pain. Irritability. Weakness. Balance problems. Funny eye movements. Tremors. Slowness of thought.

These are part of a constellation of symptoms shared by most folks who suffer from the rare disorder called Glut1.

Many parents are not familiar with the disorder and therefore don't know how to help their child when these symptoms appear. Worst of all, because some physicians can work their entire life without ever meeting a patient with Glut1, failure to diagnose the condition may be possible.

Glut1—short for Glucose Transporter Type 1 Deficiency Syndrome—is a genetic disorder impairing how glucose is transported into the brain. The result is a brain that is left constantly starving the energy it needs to grow and function properly. This is why, for most people with Glut1, fasting is accompanied by a worsening of symptoms.

Because it is so rare, seizures, cognitive delays, and balance problems present in Glut1 patients can easily be attributed to other better-known conditions, such as epilepsy.

In fact, it is possible that one percent of all people diagnosed with generalized epilepsy and 10 percent of children with absence epilepsy may actually have Glut1.

Additionally, misdiagnosis can occur because symptoms for Glut1 can be non-specific, which means they can vary depending on the particular mutation.

For young children, who are developing their brain during the first five years of their lives, undiagnosed Glut1 can have severe developmental consequences. This five-year brain development window is a crucial period where treatment could make a big difference in the prognosis of a child with Glut1. Providing the brain with the energy it needs when it's growing and developing is key to allow for the best possible outcome. However, the only way to determine if a person has Glut1 is to perform a genetic test—which finds the mutation in about 70 percent of people with Glut1—or a spinal tap.

If a child has funny spells, seizures, abnormal eye movement, developmental delay, fatigue, is very sensitive to sound, light and sensory stimulation, is irritable, and not meeting his/her milestones, that should be enough to request a Glut1 test to rule out a diagnosis. Getting an early diagnosis will help get the treatment in place to help the child grow to his/her full potential.

For more information, visit www.glut1testing.com.

— Abigail Collins, MD, Assistant Professor of Pediatrics and Neurology at the University of Colorado School of Medicine

Lilly and Roche to Collaborate on Alzheimer's Diagnostic Tool

Eli Lilly and Roche Diagnostics are partnering on the ongoing development of a commercially scalable cerebrospinal fluid (CSF) assay for amyloid-beta 1-42. Under the agreement, Lilly is responsible for certain milestone payments upon successful completion of key development objectives. Roche is responsible for the development, registration, and commercialization of the new test. "We are excited to partner with Roche Diagnostics on this important test," said Phyllis Ferrell, vice president of Lilly's Alzheimer's disease platform. "We share the same commitment to providing people with Alzheimer's disease the best care possible, which includes detection as well as diagnosis and education." Currently, healthcare providers can find evidence of amyloid in the living brain through CSF test and an amyloid brain PET scan.

Racial Demographics of Dementia Highlighted in New Study

Data from the first study to look at dementia risk in a population representing the diversity of the US revealed that the highest incidence rates were in African Americans and lowest in Asian Americans. In fact, the rate of dementia occurrence in African Americans was 65 percent higher than Asian Americans, according to the findings published in *Alzheimer's & Dementia: The Journal of the Alzheimer's Association* (February 10, 2016). The researchers used electronic health records covering medical visits over a 14-year period to identify individuals diagnosed with dementia. They also collected race and ethnicity information in the healthcare system's member database and categorized participants into the six groups. The dementia diagnoses were either Alzheimer's, vascular dementia, or non-specific dementia. The investigators found that dementia incidence

over the 14-year study period ranged from an average annual rate of 26.6 cases of dementia per 1,000 people for African Americans, and 22.2 cases per 1,000 people for American Indians/Alaskan Natives, to 15.2 cases per 1,000 people for Asian Americans. In between were Latinos and Pacific Islanders with an average annual rate of 19.6 cases per 1,000 people, and whites with 19.3 per 1,000. ■

More Headlines from NeurologyWire

Edge Therapeutics Presents Positive Data for Aneurysmal Subarachnoid Hemorrhage Agent

At the recent International Stroke Conference in Los Angeles, Edge Therapeutics, Inc. presented positive data supporting the potential pharmacoeconomic impact of its lead product candidate EG-1962 for the treatment of patients who have suffered an aneurysmal subarachnoid hemorrhage (aSAH) resulting from a ruptured brain aneurysm. In the study, EG-1962 demonstrated a 3.5 day reduction in intensive care unit length of stay (LOS) compared to oral nimodipine; median LOS was 13.5 days for EG-1962 and 17 days for oral nimodipine.

The company plans to initiate a pivotal Phase 3 study in mid-2016. **neurologywire**

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THE FDA FILE

FDA Approves Medtronic DBS for People with Parkinson's Disease with Recent Onset of Motor Complications

The FDA has approved Medtronic Deep Brain Stimulation (DBS) Therapy for use in people with Parkinson's disease of at least four years duration and with recent onset of motor complications, or motor complications of longer-standing duration that are not adequately controlled with medication.

The approval was based on data from the EARLYSTIM clinical study, which found that patients treated with Medtronic DBS Therapy and best medical therapy (BMT) reported a mean improvement of 26 percent in their disease-related quality of life at two years, compared to a one percent decline in patients treated with BMT alone.

Therapeutic Landscape for Acute Migraine Expands with New Approvals

The FDA recently approved two new sumatriptan agents—a nasal spray and an injectable—for the treatment of acute migraine in adults. ZembraceSymTouch (sumatriptan succinate, Promius Pharma) is a drug-device combination product for the treatment of acute migraine episodes, with or without aura, in adults who are inadequately managed with existing treatment regimens. It is available as a prefilled, ready-to-use, single-dose disposable autoinjector containing 3mg of sumatriptan.

Onzetra Xsail (sumatriptan nasal powder, Avanir Pharmaceuticals) is an intranasal medication delivery system consisting of a low-dose (22mg) of sumatriptan powder indicated for the acute treatment of migraine with or without aura in adults.

Synthetic Nerve Capping Device Cleared

The FDA granted 510(k) clearance for Neurocap, a device designed to reduce painful neuroma formation and facilitate tissue repair and regeneration. It is available in the US through MicroAire Surgical Instruments LLC (www.microaire.com).

Extended-Release Orally Disintegrating Amphetamine for ADHD Approved

The FDA has approved Adzenys XR-ODT (Neos Therapeutics), an extended-release orally disintegrating tablet for the treatment of ADHD in patients six years and older. It was approved via the 505(b)(2) regulatory pathway. The clinical program demonstrated that Adzenys XR-ODT is bioequivalent to a previously approved mixed amphetamine salts extended-release capsule (Adderall XR). Adzenys XR-ODT will be available in six dosage strengths, allowing healthcare providers to individualize the dose.

FDA Approves UCB's Briviact (brivaracetam) for Partial-onset Seizures of Epilepsy

UCB has received FDA approval for Briviact (brivaracetam) as adjunctive therapy in the treatment of partial-onset seizures in patients 16 years of age and older with epilepsy. Brivaracetam displays a high and selective affinity for synaptic vesicle protein 2A (SV2A) in the brain, which may contribute to the anticonvulsant effect. However, the precise mechanism of action by which Briviact exerts its anticonvulsant activity is not known.

Briviact will be available in three formulations: film-coated tablets, oral solution, and injection.

