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Triple Trouble: A Rare Case Report of PCDH19, Autism and Epilepsy in a 7-Year-Old Male Child

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ABSTRACT: Background: A 7-year-old Caucasian male presented to the emergency department with worsening aggression andbehavioral problems. He was diagnosed with seizure disorder at 9 months old and was started on Keppra. He also had delayed developmental milestones including failure to roll until 6 months, and failure to crawl until after 9 months. At 16 months he was aggressive towards other children including unprovoked attacks of biting and assault. When he was 6yrs old he underwent genetic testing which showed positive for PCDH19 mutation. The most consistent feature of this condition is the early onset ofseizures between 3 months to 3 years of age. He has been on nine anti-seizure medications since his diagnosis. For behavioral problems, he was placed on Focalin, Ritalin, Adderall patch which did not seem to work. He was then on non-stimulants, Zoloft, Prozac, Effexor, Zyprexa and Abilify all with little success.
During this admission, patient's mother described increasing acts of severe physical violence toward multiple people leading to significant bruising. He has a known past psychiatric history of ASD, ADHD,ODD and PCDH19 mutation associated epilepsy. On admission, his medications included Buspirone 5mg BID, Olanzapine 7.5mg BID, Methylphenidate 7.5mg, Ritalin 7.5mg, Zonisamide 150mg qhs, Clonidine 0.1mg qhs. He had significant sensory processing difficulties and lack of communication with peers. He was continued on Clonidine 0.1mg po qhs, Methylphenidate 7.5mg po qam and at noon, Zonisamide 150mg daily. Tapered down Olanzapine and initiated him on Risperidone. His condition improved. He was referred to Thompson center for ABA therapy. He is more interactive and has a smile.

DISCUSSION: PCDH19 gene mutations have long since been known to cause epilepsy and behavioral disturbances in females. Males, on the other hand, present as asymptomatic transmitters. PCDH19 is a gene located on Chromosome X and is responsible for the formation of a protein known as protocadherin 19. This protein is especially important as it functions as a Ca2+ dependent cell adhesion in the brain. The PCDH19 mutation, however, impedes protein formation leading to epilepsy and behavioral disturbances. Approximately 90% of symptomatic females possess the mutated gene on one of their X chromosomes. Males similarly carry the mutation on their X chromosome, however are typically asymptomatic. A rare mosaic variant of PCHD19 mutations has been seen in symptomatic males.

CONCLUSION: Although it is more difficult to think in terms of ASD if a PCDH19 patient has coexisting psychiatric comorbidities like ODD andADHD, clinicians must be keenly aware of other mood disorders. Seizures do not respond well to medications. Family education, psychopharmacological treatment, ABA and CBT were successful in treating the patient. Little is known about the long-term sequela and prognosis of PCDH19 mutation in the male patient population and thus further research is warranted.

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Unintentional Overdose on Long-Acting Injectable Risperidone

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ABSTRACT: Study Objectives: Understand the mechanism and pharmacokinetics involved in an unintentional overdose of injectableantipsychotic medication; identify how to transition between long acting injectable antipsychotics; formulate steps to ensure patient safety in an event of an unintentional overdose of injectable antipsychotic

METHOD: Review of a case study involving a 47-year-old male, with history of schizoaffective disorder and previous episodes of loss of consciousness of unknown etiology, who was admitted to the inpatient psychiatry unit at UConn Health John Dempsey Hospital for stabilization of psychotic symptoms and monitoring of unintentional overdose after initially being admitted to the ICU. He was found unconscious at home following a period of days where he self-injected four of 37.5mg Risperidone injections (estimated 150mg) along with limited adherence to prescribed oral clozapine, doxepin and lorazepam. He reported self-injecting additional medication to treat paranoia, auditory, visual and olfactory hallucinations. In the ICU, he was evaluated by Toxicology and Neurology for loss of consciousness thought to be from seizures, with no clear outcome. He was awake and alert within 24 hours of medical admission but became agitated, hostile, and psychotic prompting psychiatric admission. When his

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worsening paranoia had resulted in termination of his visiting nurse services that administered injections and in home assessments, his outpatient psychiatrist allowed him to self-administer bi-monthly injections. Over three weeks in the hospital, he was evaluated for signs and symptoms of antipsychotic overdose.

RESULTS: Initial literature review did not reveal information involving an overdose of injectable Risperidone. Thus, the time frame and symptoms to monitor were uncertain. As the injectable medication was expected to peak in 2-3 weeks and persist for 4-6 weeks, there was a concern about delayed potential side effects such as EPS, sedation, QTC prolongation and electrolytes imbalances. He was treated with oral antipsychotic medication. Clozapine and doxepin were discontinued due to patient non-adherence, side effects, and drug interactions. He exhibited signs of EPS and was started on benztropine. To simplify his regimen, he was switched to another long acting injectable, Paliperidone Palmitate, prior to his discharge.

CONCLUSIONS: Given the nature of the presentation, he was advised not to self-administer injectable medication and was referred for visiting nurse services. He was educated on the potential side effects of injectable antipsychotic medication. As there was a change in antipsychotic medications, follow up was recommended in an intensive outpatient program for psychotic symptoms and prolonged side effects. Due to the patient's concordant episode of loss of consciousness, he was advised to follow up with an outpatient long term EEG monitoring and complete Neurology evaluation.

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AXONA (Caprylidene): Medical Food Therapy For Alzheimer's Disease

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OBJECTIVES: Evaluate the current novel food therapy for Alzheimer and its adverse effects. What is the response to Axona (Capylidene) in different ethnicities and determine the generalizability of the drug use in diverse populations. What genes are linked with positive responses? What are the implications of its use in high risk populations? Its role in early detection of Alzheimer's and the arrest of the neurodegeneration in APO E4 (-) patients.

METHOD: PubMed was queried with the search terms 'Axona' OR 'Caprylidene' and the following articles were collected and reviewed.

RESULTS: Among the articles collected and reviewed, two studies extensively evaluated the safety and efficacy of using Medium Chain Triglycerides (MCT) in Alzheimer's disease. These studies genotyped patients for APO E4 status (positive/negative). According to the Reger et al. study in 2002, treatment of APOE4 (-ve) patients with MCTs reported a considerable improvement in comparison to placebo-treated patients ($P = 0.04$). The second study by Henderson et al. in 2009 demonstrated an improvement in cognitive functioning determined by Alzheimer's disease Assessment Scale- cognitive subscale (ADAS-Cog) scores in those treated with MCTs versus placebo in APOE4 -ve patients. An open label Japanese pilot study also showed improvement in cognitive functioning with Caprylidene in APOE4 (-ve) patients with Mini Mental Status Exam (MMSE) score > 14.

DISCUSSION: The FDA approved treatment options in Alzheimer's disease include acetylcholinesterase inhibitors (Rivastigmine, Donepezil and Galantamine), NMDA receptor antagonist (Memantine). These drugs only delay the progression of the disease in these patients.
MCTs are classified as medical foods, which are defined as substances that provide a specific nutritional need in a patient that cannot be satisfied by modification of a normal diet alone The FDA approved Axona as medicalfood for specific dietary management of the disease in 2009. Early metabolic changes in Alzheimer's Diseaseprior to cognitive decline and plaque deposition can possibly be prevented by early intervention with Axona, especially in high risk population (APOE4 (þ), Downs syndrome).
These trials highlight the benefits of MCT in a discrete group, and the importance of routine genomic testing in Alzheimer patients in clinical settings. A better understanding into Caprylidene's pharmacokinetics and pharmacodynamics will help us in the prevention and intervention of patients based on their genetic profiles.

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Second Generation Antipsychotics and Catatonia: A Literature Review

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