* Final Report *
Document Has Been Revised

History of Present Illness
Ibrahim is a 3-year-10-months old boy with intractable focal epilepsy likely due to genetic etiology.
Ibrahim was doing fine until the age of 3 month when he was admitted twice in Sidra because of frequent focal seizures.

His seizure semiology consists
of sudden behavioral arrest, lip smacking, tachycardia, cyanosis and oxygen desaturation. Head and eye deviation to the left or right side. Later on he started having clusters of epileptic spasms.

He responded very well to antiepileptic medications and his EEG improved a lot.
Today father reports that the child has been seizure-free for over 2.5 years.

Developmentally he is making slow progress. Currently, he can sit up and can stand with support, he reaches out for objects with either hand, he recognizes family members and he has social smile.
But he still doesn't talk and his appetite and weight gain has been poor.

Current antiepileptic medications:
1- lacosamide 15 mg twice a day (2.2 mg/kg/day) **(seizures improved significantly after adding this medicine).**

Previous antiepileptic medications:
Clobazam, phenytoin, oxcarbazepine.

Pevious workup:
- The most recent EEG in November 2020 showed **significant improvement** with no epileptiform discharges, he has only mild slowing of the background activity.
- **MRI was unremarkable.**
- metabolic work up was unremarkable.
- **Positive Family History of epilepsy in several family members however father does not have any further details.**
- **Whole exome sequencing** revealed variants in 2 different genes:
  1- non currently described gene PRKACB gene de novo heterozygous variant described in multiple congenital anomalies.
  2- ZFYVE26 gene compound heterozygous VUS known to cause AR spastic paraplagia CP type 15, parents are each heterozygous for the same variant.

Review of Systems
Constitutional: [No fevers, chills, sweats]
Eye: [No recent visual problems]
ENMT: [No ear pain, nasal congestion, sore throat]
Respiratory: [No shortness of breath, cough]
Cardiovascular: [No Chest pain, palpitations, syncope]
Gastrointestinal: [No nausea, vomiting, diarrhea]
Genitourinary: [No hematuria]

Physical Exam
T: 36.6 °C (Oral) **SpO2:** 96% **WT:** 13 kg
General: He looks well hydrated, well-nourished with no apparent distress.

Dr. Husam Kayyali
A. Senior Medical Staff
Pediatric Neurology
QCHP License: P12183
Sidra Medicine

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HEENT: normocephalic, fontanels are soft, no dysmorphic features and no signs of trauma.
Abdomen is soft, no masses and no organomegaly.
Skin: bilateral polydactely.
Neurological: he can sit up with some support, he reaches out for objects with either hand, he recognizes family members and he has social smile. He crawls but he can't stand or walk.

Assessment/Plan

Focal epilepsy

Ibrahim is a 3-year-10-months old boy with epilepsy likely due to genetic etiology.
His seizures were difficult to control initially then he became seizure free and maintained good seizure control for the last 2.5 years on lacosamide. Then in July 2022 we weaned him off Keppra and he remained seizure-free.

Today I recommend to continue with lacosamide 15 mg twice a day.
For the hyperactive and aggressive behavior we will treat him with clonidine 0.01 mg 3 times a day.
Follow up EEG.
we will see him in the clinic for follow-up after 4 months.

The child has severe Developmental disabilities and he is totally dependent on his parents for all activities of daily living. I support the parents request to have disability car parking permit.

Also considering the child's medical condition, he is at risk for decreased bone density and it is essential for him to have enough sun exposure for his bone health. All help extended to his parents to have proper accommodation is highly appreciated.

Signature Line
Husam Kayyali

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Completed Action List:

* Perform by Kayyali, Husam (ID 25201) on 12 03, 2023 16:52 AST
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