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Pediatric Neurology Second Opinion

Extended Report

Patient:

DATE: 04/4/2022

Discussion, analysis and recommendations:

_____ is a 23 month old male ex-37 week male with developmental delay most pronounced in the realm of expressive language. He has a history of failure to thrive and manifests severe oral aversions. By history, he has both hypotonia and some increased appendicular tone. There is concern for cortical fisting bilaterally and dragging of the left leg for which he receives both occupational and physical therapy. He positions his hands and fingers in unusual positions. Pregnancy was remarkable for threatened pre-term labor necessitating bed rest, terbutatine, and progesterone injections. Pregnancy was complicated by gestational diabetes and pre-eclampsia requiring treatment with Nifedipine (Procardia). Birth history is remarkable for intermittent fetal decelerations. _____ was born at 37 weeks weeks by spontaneous vaginal delivery. Birth weight and Apgars are unknown but he breathed spontaneously and had an uneventful neonatal course. He was discharged one day early from the hospital. His motor milestones are not provided but he is reported to have been developing close to age appropriate until 17 months of age. There does not appear to be developmental regression but rather failure to appropriately acquire both motor and speech milestones. Currently, Caden is about 20 pounds, placing him well below the 2nd percentile for age. He has not gained any weight in the last 8-9 months. From a speech standpoint, he has only 5-6 words and the majority of his speech is unclear.

MRI from 2-19-2014 indicates no structural abnormalities though no comment is made of white matter myelination which would be of interest.

Review of video provided by mother shows a thin toddler with wide based gait that is at times unsteady. Occasional head nodding is noted. He self feeds using a fork. He has a facial diplegia

suggesting poor oral-motor tone. He maintains his wrists in flexion and has a tendency to fist. No overt facial dysmorphisms are appreciated though he has a prominent forehead.

In summary, _____ is a 23 month old male born at 37 weeks' gestation with history of developmental delay, failure to thrive, severe oral aversion, profound speech delay and stereotyped interests. This phenotype can be seen in a number of neurogenetic syndromes, metabolic disorders and mitochondrial disorders. Further work up as follows is recommended:

1) Fragile X, Microarray

2) Ensure normal newborn screen

3) Comprehensive metabolic work up to include: plasma amino acids, urine organic acids, lactic acid, pyruvic acid, ammonia, and acylcarnitine profile

4) EEG to evaluate for epileptic encephalopathy

5) Evaluation by a developmental pediatrician

I do not believe sinus venous thrombosis or stroke would explain all of this child's difficulties. He would benefit from a thorough neurogenetic evaluation as can be done at Medical Neurogenetics in Atlanta, GA.

Dr. requested a consultation from Dr. Board Certified Pediatric Neurologist -Electronically Signed by:

on 04/01 /2014 7:45 AM