Mauritius, 29 June 2022

Patient : Ayesha AMEERKHAN (F) born 25 March 2022

Dear Doctor,

I would like to refer you the case of the above named patient who has hypotonia, dysmorphic features and recurrent episodes of aspiration. We do not have yet a precise diagnosis of her underlying neurological condition.

Familial history :
- No parental consanguinity.
- Father's cousin has a son with milestone retardation (psycho and motor)

- Sibling passed away. Born at 37 weeks. BW 2.55 kg. NVD. Immediate cry. Excessive cry,hypotonia, microcephaly, high trypsine level on Guthrie test. absent corpus callosum and dilated ventricles. Child passed away with septic shock at the age of 50 days. Cultures all negative though : blood CSF negative as well

Antenatal and peri-natal history
- Pregnancy uneventful. Mother does not consume alcohol or drugs. No medication except vitamins
- Born by ventouse vaginal delivery. Term 38 weeks. Apgar 9-9-10. Transient tachypnea of new born. Clinically, child as high arch palate and also cracked lips. No cleft lip of cleft palate. No hypoglycemia. Blood tests done not in favor of sepsis. Cultures negative

Current issues
As from the age of 3 months, child has had the
- Three admissions for respiratory distress, cough needing oxygen. RSV (-), Covid (-), Inflenza (-). The child is becoming more and more sleepy and lethargic. The respiratory issues of the last admissions were due to aspiration while swallowing, not during reflux.
- Child is sleeping for long periods and less reactive and lethargic.
- Loss of certain psychomotor acquisitions like movement of head sideways. Child was lifting her head but has now stopped with a complete head lag. No more eye contact and focusing.
- Microcephaly : OFC < P3 but remains paralell to Percentile 0.4
- Loss of weight.
- No abnormal movements that would suggest an epilepsy. No infantile spasm, no clonus, no myoclonus.

Clinically,
- Neurological : she does not look in the eyes, is hypotonic. She is poorly reactive. High arch palate. Her reflexes are present. AF open. Head lag ++.
- Cardiac : S1S2 no murmur. Extremities warm and pink. CRT 2 sec. Fem pulses present.
- Breathing : Air entry present with crepitations. No recession except on admission. Snoring with significant upper airway obstructions requiring suction almost every hour. Her SpO2 is

corrected after upper airway clearance. Oxygen required at admission. No more needs once child is keep Nil by mouth.
- Abdomen : Not distended. No HSM. BS ok.

Tests done
- Abdominal ultrasound : Normal. No hepatosplenomegaly
- EEG : Suggestive of encephalopathy. The baseline rhythm consist of slow delta and theta waves of high amplitude over all leads. No epileptic discharge noted. Artifacts secondary to muscular movements.

- Cardiac echo in maternity : Small ASD. Stretched PFO.
- Cranial ultrasound : Normal
- Chest Xray : normal
- MRI brain : not done yet. That will be conducted once child is more stable

- PCR for RSV, COVID and influenza : negative during all 3 admissions in Paediatric Ward
- Blood gas : no metabolic acidosis.
- Urine : no ketones.
- Blood tests :

- Next Generation Sequencing Clinical exome sent : awaiting results
- Guthrie test : normal

SUMMARY
- Unknown underlying neurological condition
- Positive familial history.
- Regressive psychomotor skills : degenerative ? Onset around 3 months of age
- Dysmorphic features + microcephaly + swallowing difficulty + recurrent aspiration

We would like to refer you the case in order to continue the work up and reach a diagnosis. I remain available for any more information required. There is a link and a QR code which leads to a folder with her medical file.

Kind regards

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SCAN ME

Password for Link below : AA25032022
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