

GBN BVN Autumn Meeting 2023



The NICU, not always a “cool club”

S. VERBEECK

R. CILIO, E. CARAPANCEA, K. CARKEEK

History

Setting the scene

Cliniques Universitaires Saint Luc

Just when everything quietening down one evening

21h

Telephone call from referral hospital



History

But... 😞

Baby girl born 3 hours prior

No relevant maternal history
G1P0
O positive
Induction for pre-eclampsia
ROM <18h
Vaginal delivery

Shoulder dystocia
Apgars 2-5-8
Ventilation 1 minute
Hospitalization with CPAP

What else?

CPAP PEEP 6 Fio2 60 > 21%

Fluid bolus 10cc/kg at H2 for hypotension

Antibiotic had been started (amoxicilline – amukin)

Glucose bolus 2cc/kg for hypoglycemia followed by peripheral perfusion with G10%

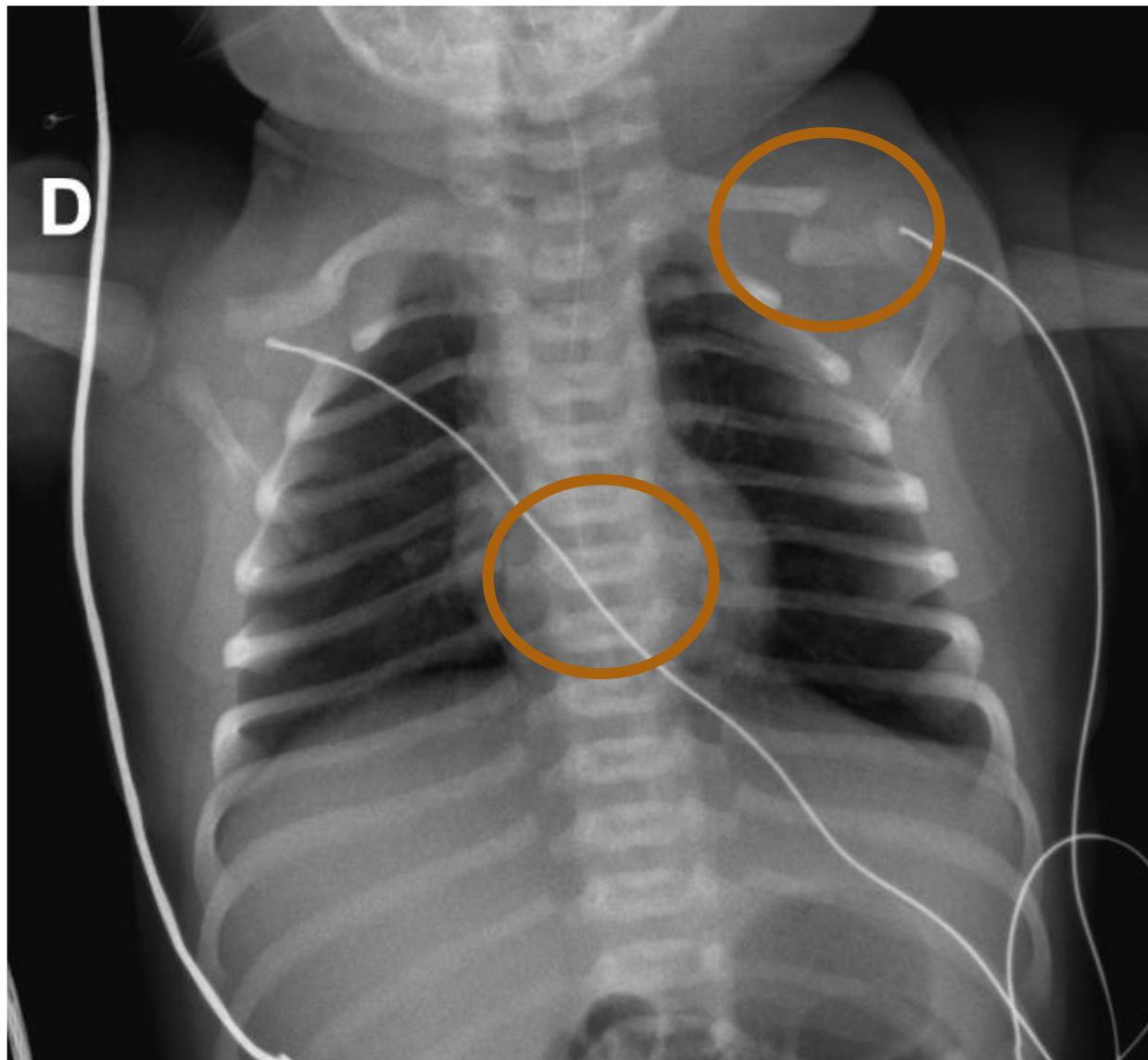


Blood gas evolution

Umbilical cord: pH 7,1 – lactate?

H1: pH 7,13 – lactate 10,36 – base excess -12

H2: pH 7,29 – lactate 4,9 – base excess -3,7



Chest Radiograph

What do you see?

Reason for the call?

“ We thought we remembered something from a Saint-Luc lecture about “mild asphyxia” cases and to always discuss for possible transfer, monitoring and possible cooling?”



“Could you come and have a look? “

Evaluation on arrival at Saint-Luc

Clinical examination

Discrete axial hypotonia

Further neurological exam normal

Sarnat 0

Thompson 2

What would you do next?

Evaluation lead to investigations

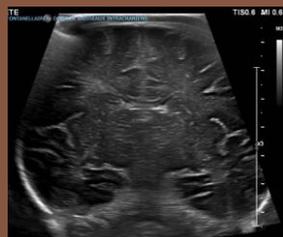
Clinical examination

Discrete axial hypotonia
Further neurological exam normal
Sarnat 0
Thompson 2



Head Ultrasound

Normal



EEG + CFM

Evaluation lead to investigations

Clinical examination

Discrete axial hypotonia
Further neurological exam normal
Sarnat 0
Thompson 2



Head Ultrasound

Normal



EEG + CFM

Normal background

So... what do we have...

(Mild) Asphyxia without encephalopathy

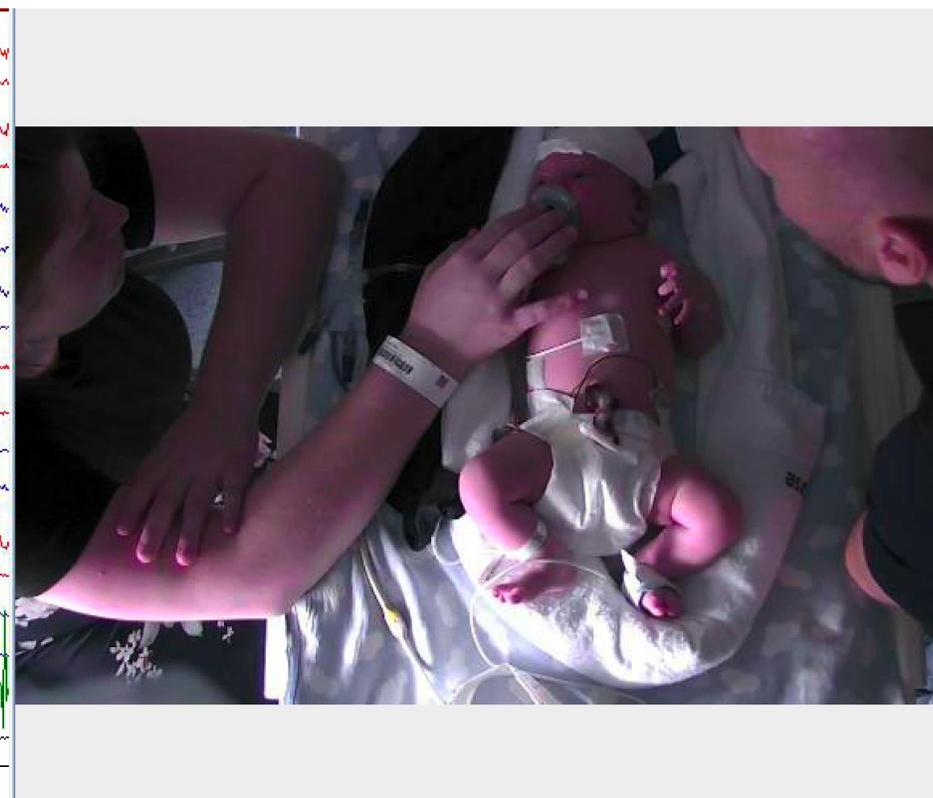
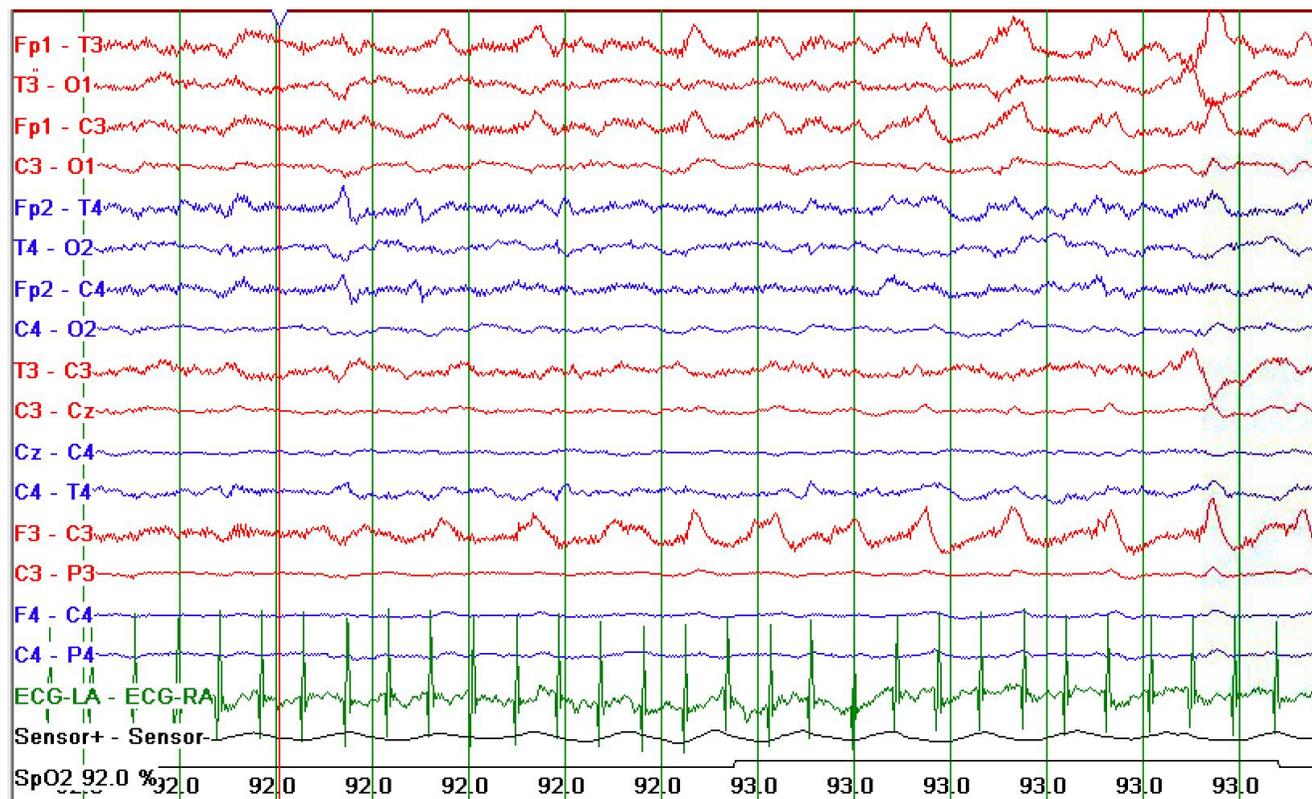


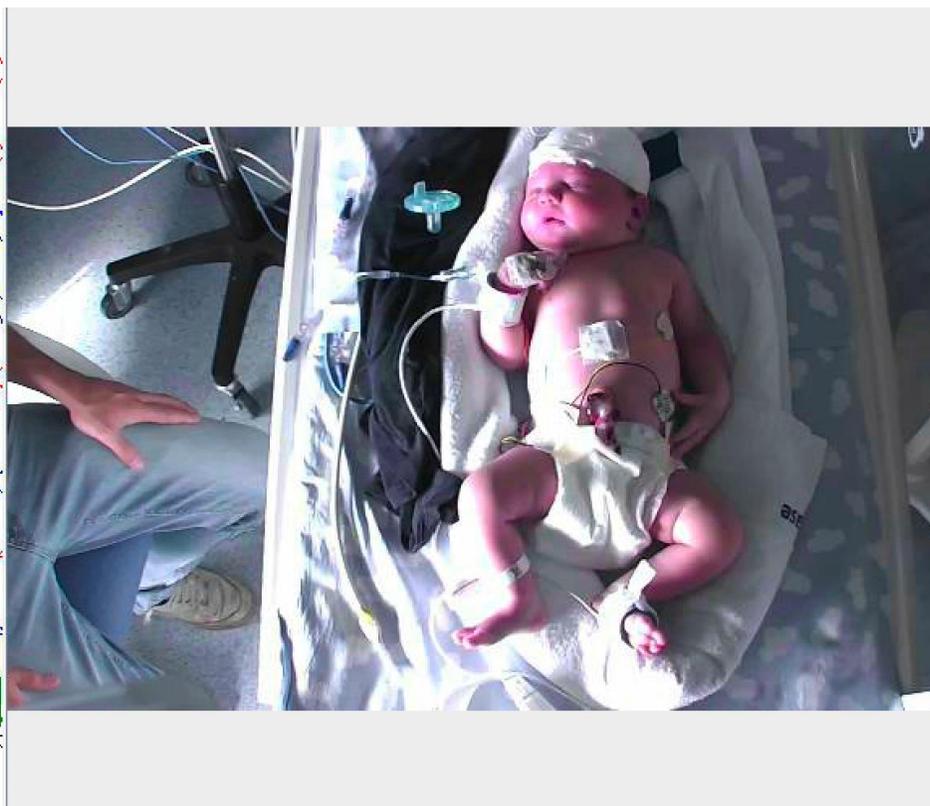
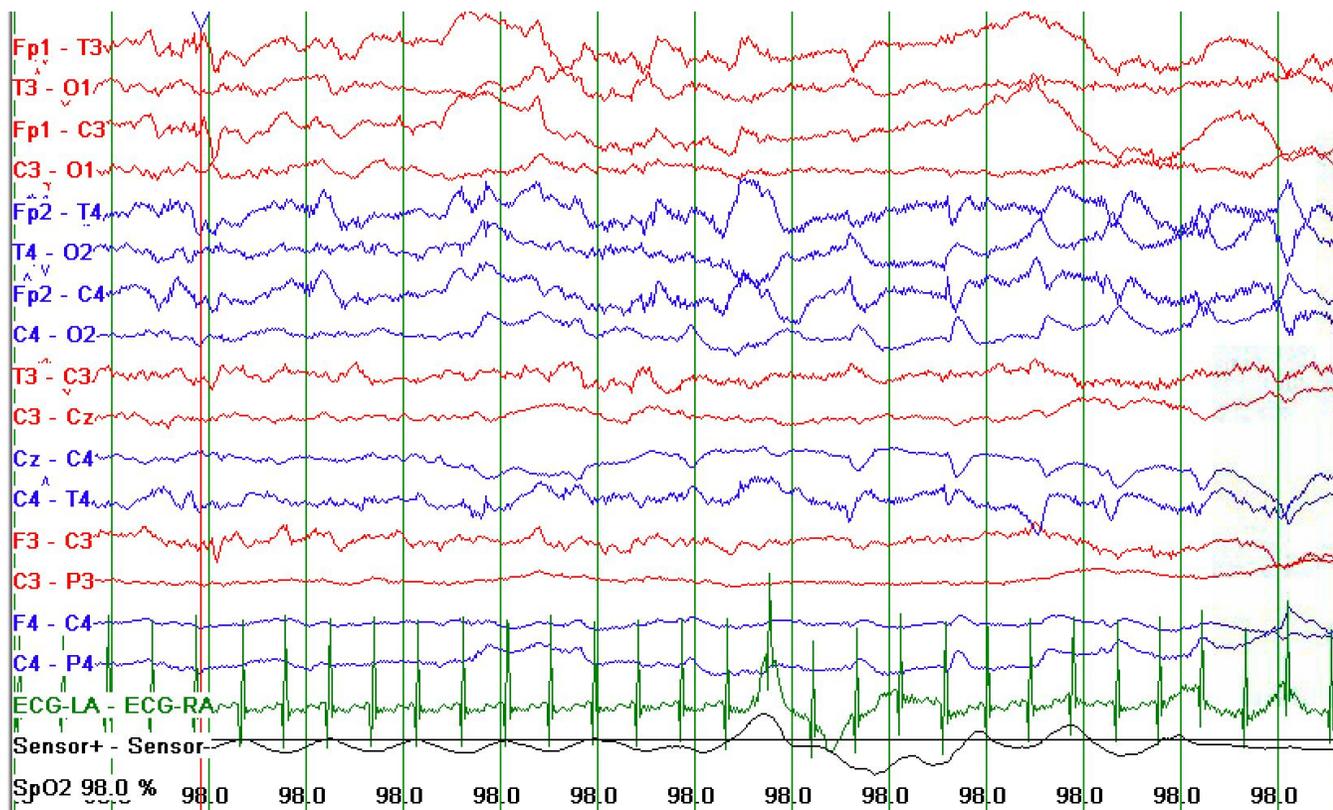
Criteria for cooling?

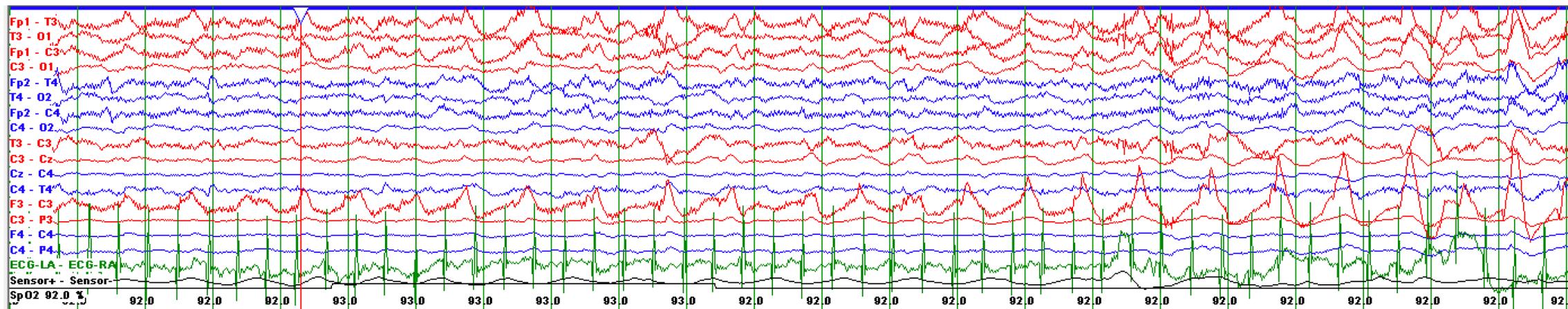


Further observation

But... at 24h of life

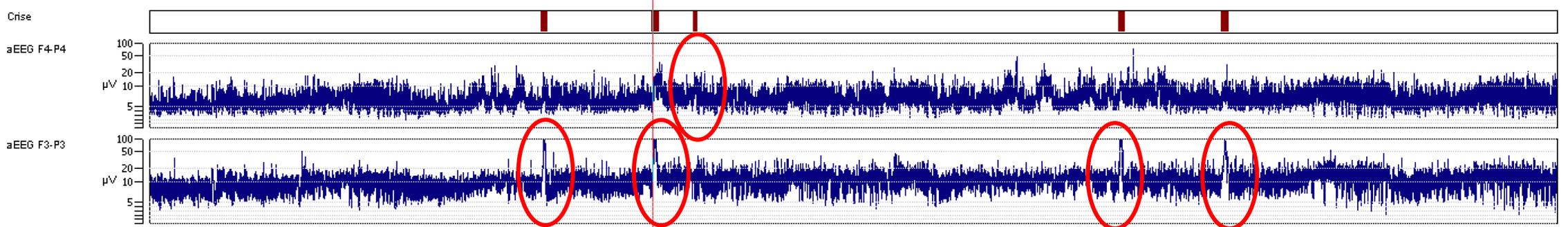






Temps écoulé (h:m:s) 04:02:25 04:19:05 04:35:45 04:52:25 05:09:05 05:25:45 05:42:25 05:59:05 06:15:45 06:32:25 06:49:05 07:05:45 07:22:25 07:39:05 07:55:45 08:12:25 08:29:05 08:45:45 09:02:25 09:19:05 09:35:15

Heure du jour (h:m:s) 14:00:55 14:17:35 14:34:15 14:50:55 15:07:35 15:24:15 15:40:55 15:57:35 16:14:15 16:30:55 16:47:35 17:04:15 17:20:55 17:37:35 17:54:15 18:10:55 18:27:35 18:44:15 19:00:55 19:17:35 19:34:15





So in summary...

Piecing the puzzle together

5 episodes of focal seizure activity

4 left and 1 right sided, bilateral independent

Associated with uncoordinated movements, contralateral myoclonia, crying

What would you do?

Arguments for seizures secondary to asphyxia?



What about these seizures?

No clinical encephalopathy

Normal initial background EEG

Atypical timing of seizures for asphyxia

Atypical aspect of seizures

- Short in duration
- Depression following seizure

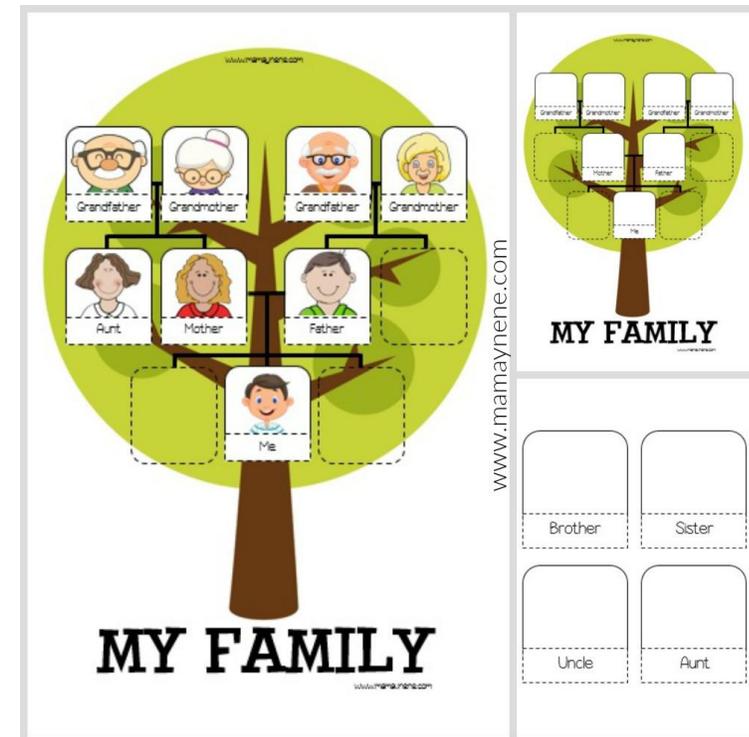
What do you think?

-> Could this be neonatal onset epilepsy?



An assistant asked further into the family history!

-> Cousin with childhood epilepsy at 4 months of age



Treatment and follow up

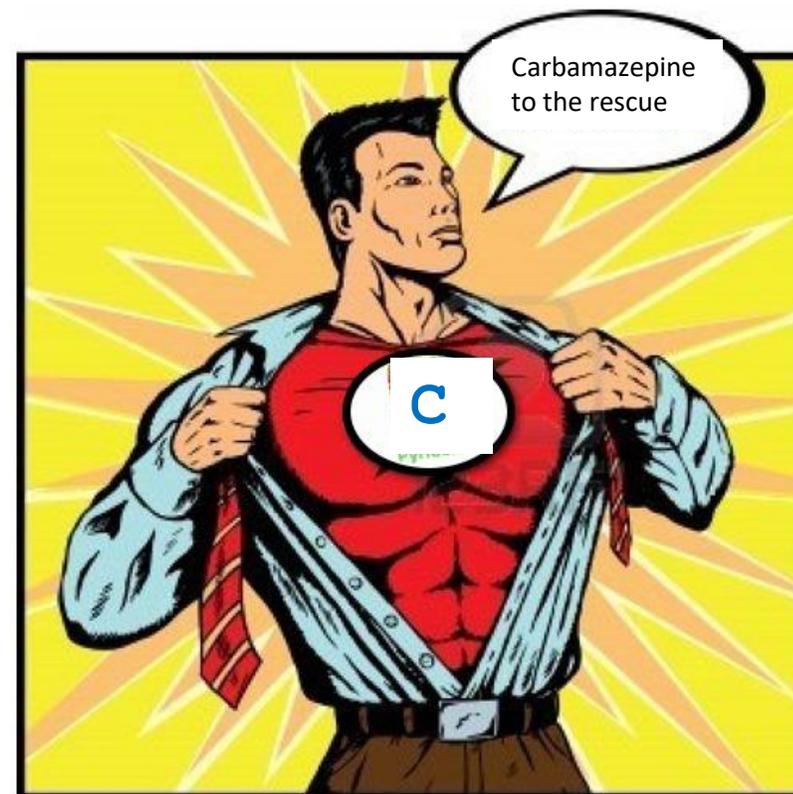
Carbamazepine started

->No more seizures after first dose of carbamazepam

Blood for Genetic panel sent

->Confirmation: SCN3A mutation

Now 3 months of age, doing well.



SCN3A

Voltage gated sodium channel > gain of function mutation

Neonatal-onset epilepsy

75% developmental delay

75% cortical malformation on MRI



Take home messages

Importance of referral

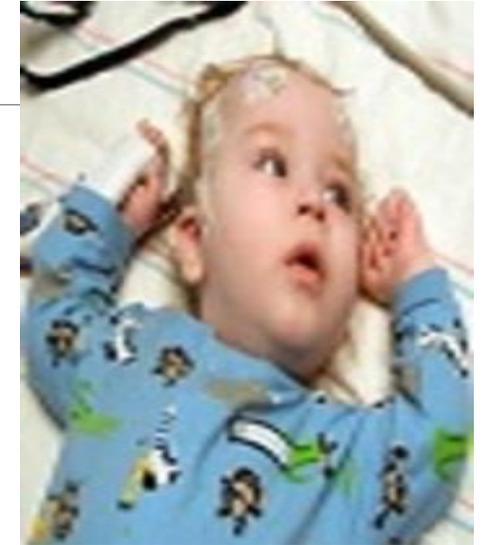
Importance of continuous video EEG monitoring and NOT just short EEGs

If the puzzle piece does “not fit” → usually another image/puzzle!



Multidisciplinary team work → Neonatal, Neuro, Genetics

Advances in genetic diagnoses and rare diseases



References

Carapancea E, Cilio MR. A novel approach to seizures in neonates. *Eur J Paediatr Neurol*. 2023 Sep;46:89-97.

Pressler RM, Lagae L. Why we urgently need improved seizure and epilepsy therapies for children and neonates. *Neuropharmacology*. 2020 Jun 15;170:107854.

Zaman T, Helbig KL, Clatot J et al. SCN3A-Related Neurodevelopmental Disorder: A Spectrum of Epilepsy and Brain Malformation. *Ann Neurol*. 2020 Aug;88(2):348-362.