A Case of Rhombencephalosynapsis

Dr. Midhun Ramesh, DNB Resident
Department of Pediatrics
Southern Railway Headquarter Hospital
Ayanavaram, Chennai-23
Particulars of the patient

- Age: 2.5 yrs
- Sex: female
- DoB: 19th Nov 2008
- Resident of Erode dst T.N

Presenting complaints
- Delayed attainment of mile stones
H/O Present illness

- 3rd Child
- 3rd degree consanguinous marriage
- Uneventful antenatal period at 1st & 2nd trimester
- Had a fall at 3rd trimester which precipitated labor
- Born at 34 weeks of GA; NVD
- Birth Wt- 1.75 kg
- Cried immediately after birth
- Neonatal period uneventful
H/O Present illness

- Mother noticed for the first time at 8 months of age that the baby is not attaining milestones like her elder siblings.
- Taken to family doctor
- Told to have some neurological illness/ Mental Rtd
H/O Present illness

- Child is gradually but slowly attaining milestones over last 1 year
- Now she walks without support, can speak 2/3 bisyllables, feeds with spoon from cup (with spillage), enjoys music & watches T.V
- No h/o loss of acquired milestones
- H/o febrile convulsions at 1yr of age
- Immunized uptodate
Family History

34 yrs

3 degree

26 yrs

9 yr

7 yrs

2 yrs
General exam

- Alert, playful & co-operative
- Vitals - Nrl

Anthropometry
- wt - 9.9 kg
- Ht - 90 cms
- HC - 43 cms (microcephaly)
- MAC - 14.5 cms
Examination of CNS

**Higher mental function**
- Interested in surroundings & oriented to mother.
- Lt handedness
- Speech - speaks bisyllables
- Primitive reflexes - absent
CNS

- CN- 3,4,6- deviation of right eye to temporal side in neutral position. **Non paralytic squint of right eye**
- Motor system
- No atrophy/hypertrophy of any muscles
- No foot drop/wrist drop
- Bulk- normal

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CNS

- Grade 5 power in all limbs
- Reflexes
  
  Superficial reflexes all present

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CNS

- No cerebellar signs
- No meningeal signs
- Gait- child walks with wide based gait
- Spine – normal
- Other systems- NAD
Summary

- 2.5 yr old female child 3rd product of a 3rd degree consanguineous marriage born at 34 wks of GA, with uneventful neonatal period presented with global developmental delay microcephaly, nonparalytic squint of Rt eye, hypertonia of rt limbs, upgoing plantar on Rt and absent cerebellar signs.
Clinical diagnosis

- Rt hemiplegic Cerebral palsy
- Microcephaly
- Global develop delay
MRI

- Olivopontocerebellar atrophy
- Atrophy of cerebellar vermis
- Atrophy of medial cerebellar hemispheres
- Fusion of superior cerebellar peduncles
- Dialated fourth ventricle
Rhombencephalosynapsis (RES)

- RES is a rare congenital malformation of posterior fossa due to primary failure of vermian differentiation.
- RES is characterized by agenesis/hypogenesis of the cerebellar vermis.
- All published cases have been sporadic.

- **Lmx1a** gene mapping to 1q21-q23 regulate early developmental events at the pontomesencephalic junction.
- Molecular analysis may reveal a mutation in the Lmx1a gene unique to RES.
Discussion

- First case in the literature was described by Obersteiner in 1914, in a post-mortem of a 28-year-old man who died by suicide.

- DeMorsier named the disorder "rhombocephalosynapsis"

- Gross and Hoff in 1959 amended it to "rhombencephalosynapsis"
RES is a vermian maldevelopment syndrome, but unlike other vermian maldevelopment syndromes, such as Dandy-Walker malformation or Joubert syndrome or tectocerebellar dysraphia there is no disconnection of the cerebellar hemispheres.

The pathogenesis of this midline defective malformation remains unclear and causative factors are controversial.

- The current teaching is that its related to a disturbed development of the cerebellum between 28 and 41 days of gestation.
Rhombencephalosynapsis diagnosed in childhood: Clinical and MRI findings By Jalel Chemlia, Mejdi Abrouga, Kalthoum Tlilib, Abdelaziz Harbia

- The child had spastic diplegia, facial dysmorphia (low set ears and hypertelorism) and normal intellectual development (normalspeech). Neurological examination revealed spastic diplegia, lower limb hyper-reflexia and bilateral sign of Babinski.
Case 1:
A 20-year-old woman presented with complaint of depression after her father's death.
Her personal medical history was unremarkable
Her neurological exam was normal
She had normal intelligence
Case 2

- A 14-year-old girl presented with complaint of involuntary movements (tics) of long evolution.
- Neurological exam was normal
- There were no other complaints at that time.
- She had normal intelligence
Largest series described so far (nine patients)
Clinical findings ranged from mild truncal ataxia and normal cognitive abilities to severe cerebral palsy and mental retardation
Clinical findings may varied according to posterior fossa pathology & associated supratentorial anomalies
Three cases with rare complex midline malformations of the CNS, diagnosed prenatally by fetal MRI.

- Two cases revealed holoprosencephaly;
- One case demonstrated rhombencephalosynapsis during evaluation for hydrocephalus
Discussion

- No specific clinical picture has been associated with rhombencephalosynapsis.
- There was no correlation between the neuroimaging findings and the clinical manifestations.
- Although behavioral and intellectual impairment is generally reported in rhombencephalosynapsis, it’s not inevitably associated.
- The clinical presentation is variable ranging from early death to variable degrees of cerebellar dysfunction and developmental delay.
- Clinical presentation and prognosis are extremely variable and generally depends on the associated supratentorial anomalies.
Discussion

- There are less than 50 cases in literature & great majority of these cases belongs to pediatric age group.
- Nineteen cases have been reported in the pre-MRI era, and all the cases were detected at necropsy. One case with CT & Rest of the cases was diagnosed by MRI.

Associated findings on MRI include:
- Deficiency or absence of the septum pellucidum,
- Dysgenesis of the corpus callosum and the anterior commisure,
Discussion

- Fused fornices and thalami Superior and middle cerebellar peduncles, along with dentate nuclei and inferior colliculi, are generally fused, giving rise to the typical diamond-shaped fourth ventricle on axial scans.
- A case report stating association of RES with single umbilical artery in IJP.
- **Gomez Lopez Hernandez Syndrome** is the only syndrome associated with RS at this time. Gomez-Lopez-Hernandez syndrome consists of RS, alopecia, and trigeminal anesthesia.
As a conclusion,

- Although a rare malformation, rhombencephalosynapsis should be considered in the differential diagnosis of the cerebellar malformations.

- This case is presented to sensitize Pediatricians about the clinical entity of RES.