SUBEPENDYMAL GIANT CELL ASTROCYTOMA (SEGA) IN A NEONATE WITH MULTIPLE CARDIAC RHABDOMYOMA

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INTRODUCTION

- Tuberous Sclerosis Complex (TSC) is a genetic neurocutaneous disorder affecting the brain, kidneys, skin, heart, eyes, lung, and liver.
- Perinatally detected TSC lesions commonly involve the heart and the brain. Cardiac rhabdomyoma (CR) is a common presentation on routine prenatal ultrasound (US)
- Congenital SEGAs are seen in 2.2% in the first 3 months of life [3], with only 27 published cases diagnosed before 12 months, and majority having rapid growth and poor prognoses
- We present a newborn with prenatally detected CR manifesting as multiple intracardiac masses and congenital SEGA noted on postnatal neuroimaging studies.

OBJECTIVES

- To present a case of neonatal Subependymal Giant Cell Astrocytoma in TSC.
- To discuss the incidence of congenital SEGA, its manifestations, diagnosis, management, and prognosis.

CASE

- A 30-year-old G3P2 (2002) mother routine US at 27 weeks age of gestation (AOG) showed fetal cardiomegaly and hyperechogenic focus on the left ventricle. Fetal 2D-echocardiography showed multiple intracardiac masses with mild right ventricular outflow obstruction and a small ventriculo-septal defect.
- The patient was delivered at 37 weeks AOG via normal spontaneous delivery, birth weight 3080 grams (p50), head circumference 33.5 centimeters (p50), and APGAR score of 7,9. Physical exam showed a grade 2 holosystolic murmur, 2nd left intercostal space, parasternal area with good perfusion. Neurologic exam was unremarkable. Ophthalmologic exam revealed an isolated 2x2 mm nodular mass inferior to the right optic nerve, most likely a retinal astrocytic hamartoma.
- Laboratories:
  - 2D-echocardiography: patent foramen ovale with multiple homogenous non-obstructive masses (Fig 1), and 1.2x1.8cm mass in the right ventricular outflow tract
  - Cranial MRI: 2.9 x 1.6 x 1.7 cm SEGA (Fig. 2a) along the right posterior horn of the lateral ventricle, multiple cortical and subcortical tubers (Fig. 2b), radial bands, SENs along the lateral ventricular wall, and bifrontal hamartomas
  - EEG: electrographic discharges without clinical correlates at right parietal, occipital, posterior temporal areas
- Subsequent follow up showed:
  - Hypopigmented macules at 1 month, with mild developmental delay
  - Stable intracranial lesions on repeat neuroimaging at 3 months old
  - Infantile spasms at 6 months old controlled with high dose prednisolone
- She had occupational and physical therapy sessions with regular surveillance and follow-up with multi-disciplinary team

DISCUSSION

Most congenital SEGAs have 6-fold higher growth rate than non-congenital SEGA, particularly in the 1st 6 months of life. Surgical management in these young infants is challenging, with poor surgical outcome. Despite the lack of long term safety data and FDA approval for <1 year of age, mTOR inhibitors have been used in neonates and infants as primary treatment for clinically significant congenital SEGA unsuitable for surgical intervention or as add on treatment for partially resected tumors without major detrimental reactions. Mortality rate of congenital SEGA is almost 80%.

CONCLUSION

Congenital SEGA is an extremely rare occurrence in TSC that is mostly associated with rapid growth and poor prognosis, but a benign course and slow growth have been reported, like the case presented. A neonate presenting with CR should make one highly suspect for TSC to establish early diagnosis, prompt management and close surveillance to prevent or mitigate its debilitating consequences. Given the hereditary nature of the disease, it is prudent to perform genetic testing and counseling in these patients, which is a limitation of this study in the local setting.

IMAGING RESULTS

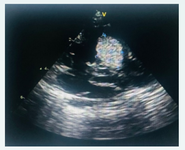

Fig 1. A 1.2x1.8cm mass in the right ventricular outflow tract

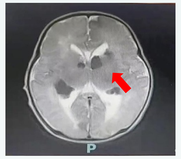

Fig 2a. A 2.9 x 1.6 x 1.7 cm SEGA at the right posterior horn of the lateral ventricle

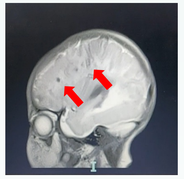

Fig 2b. Multiple cortical/subcortical tubers in the cerebral white matter and multiple radial bands

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