MSD Publications
Research is creating new knowledge

MSD Global Journal of Clinical and Medical Case Reports

Case Report

Received: 08 November, 2024
Accepted: 20 November, 2024
Published: 21 November, 2024

***Corresponding author:** Livrinova Vesna, University Clinic for Obstetrics and Gynecology in Skopje, Faculty of Medicine, Ss. Cyril and Methodius University in Skopje (UKIM), North Macedonia; Email: livrinovav@gmail.com

Copyright: © 2024 Livrinova V, et al. This is an open-access article distributed under the terms of the Creative Commons Attribution License, which permits unrestricted use, distribution, and reproduction in any medium, provided the original author and source are credited.

The vein of Galen aneurysmal malformation associated with Turner syndrome mosaicism -a case report

Livrinova Vesna[1]*, Elena Shukarova[2], Irena Todorovska[1], Slagjana Simeonova Krstevska[1], Sanja Ristovska[1], Renata Dimitrovska[1], Orhidea Stomnaroska[1]

[1]University clinic for obstetrics and gynecology in Skopje, Faculty of Medicine, Ss. Cyril and Methodius University in Skopje (UKIM), North Macedonia.

[2]University Clinic for Pediatric Diseases, North Macedonia, Ss. Cyril and Methodius University in Skopje (UKIM).

Abstract

Prenatal diagnosis of the vein of Galen aneurysmal malformation. Congenital malformation develops during weeks 6-11 of fetal development as a persistent embryonic prosencephalic vein of Markowski. The incidence of this malformation is estimated to be high. The true incidence is unknown but estimated to be 1 in 10,000 to 1 in 25,000 births. malformation accounts for 30% of pediatric congenital vascular malformations and about 1% of all pediatric congenital anomalies. Infants often die of high-output congestive heart failure [1,2]. The presenting case report's main aim is to illustrate the disease with ultrasound performed during pregnancy and Magnetic resonance characteristics, postnatal evaluation, parenteral counseling, and their decision for further treatment in the neighboring country [3]. It should be stressed that the fetus has mosaicism of Turner Syndrome, two lines, normal and unusual abnormal form. The pregnancy was 34 gestational weeks with intrauterine growth restriction. The limitation in the precision of prenatal diagnosis at primary care obstetricians and the use of additional diagnostics tools for improving the diagnosis can lead to unwanted events of pregnancy, like intrauterine fetal demise. This does not happen in this case.

Case Report

Case Presentation: A 30-year-old multiparous woman (G2.P1) presented to the clinic with a second pregnancy because of intrauterine growth restriction below the fifth percentile and the presence of oligohydramnios. She had 10 antenatal checkups before she was referred to our clinic for second-trimester screening for congenital abnormalities as normal pregnancy during checkups. Her results of PRSCA 1 show a high risk for aneuploidies, but she refuses further prenatal diagnosis for aneuploidies at the clinic, ultrasound assessment was done, and we found intracerebral Midline hypoechogenic vessel with turbulent flow, cardiomegaly, shorter long bone for 3-4 week, micrognathia. (See ultrasound pictures in Appendix) CT angiography and MRI have confirmed the malformation (Pictures 1 and 2).

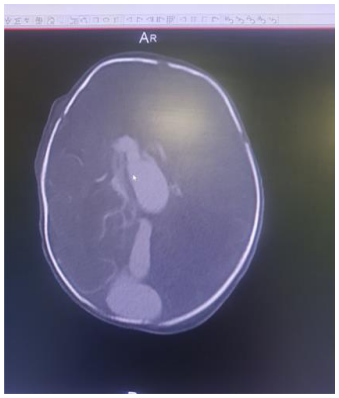
Citation: Vesna L, Shukarova E, Todorovska I, Krstevska SS, Ristovska S, et al. (2024) The vein of Galen aneurysmal malformation associated with Turner syndrome mosaicism -a case report. MSD Glob J of Clin med Case Rep. 2(1): 006-008. DOI: 10.37179/msdgjcmcr.000005

She was dilatated already 3-4 cm, she delivered spontaneously the next day. She delivered a live female fetus weighing 1840 g with 44 cm, AS 7/8. After delivery, the investigation was done. The vein Galen malformation was proved. some dysmorphic signs were present- the newborn occupies the so-called frog position. Slanted and narrow eyelids. Round face. Micrognathia. broad forehead with prominent tubercles. blue sclera. wide hairline on the neck, short neck. Low-set auricles with (incompletely turned ear helixes) untwisted helices Elevated philtrum with CUP Shape mouth, with drooping corners of the mouth. (Picture 3). Karyotype from leukocyte staining with GBT, from the blood of a newborn 80% 46, x.psu.idic (Xq),20% 45, XO. An iso Xq chromosome that has successively undergone deletions, and inversions and appears as pseudo-isocentric. The same was confirmed by karyotyping software Icarus Meta Systems (Pictures 4 and 5).

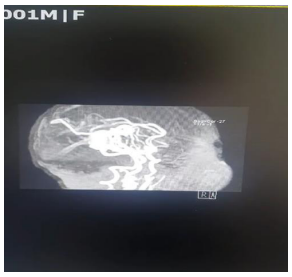
Conclusion

Turner syndrome (TS) is one of the most common sex chromosome abnormalities and results from total or partial monosomy of the X chromosome. It occurs in 1 in 2000 newborn girls and is also believed to be present in a larger proportion of conceptuses. Various anatomic anomalies have been associated with TS and the consequences of late recognition of these anomalies can be devastating [4]. Cardiovascular abnormalities are usual for this syndrome, but the Vein of Galein malformation was not connected to this syndrome. In fact, in the same fetus, two abnormalities were present. Other anatomic variations increase morbidity in this population and negatively impact the social and reproductive aspects of their lives, like neurological, genitourinary, otolaryngologic, craniofacial, and skeletal defects associated with TS [5, 6]. After postnatal evaluation and parenteral counseling, they decided to further treatment in a neighboring country for cauterization of vascular cerebral abnormality (Pictures 1 and 6).

Conflict of interest: No declared.

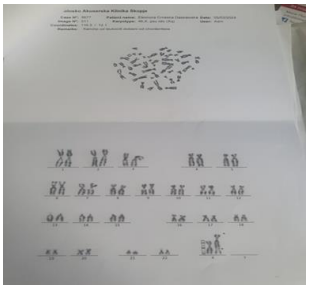


Picture 1: Picture 1 MRI of the head.

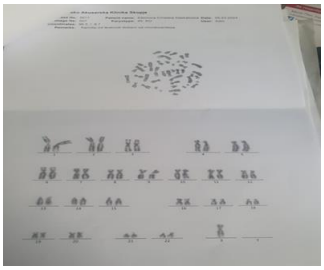


Picture 2: Picture 2 KT angiography of newborn head.

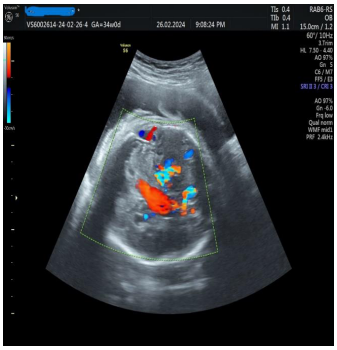
Picture 3: Picture 3 Neonatal Dysmorphia.



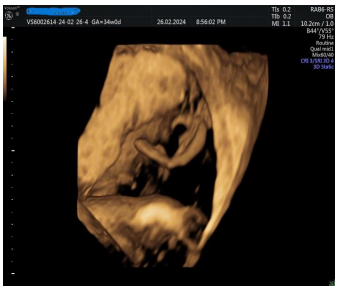
Picture 4: 46, x.psu.idic (Xq) It was confirmed with microarray.



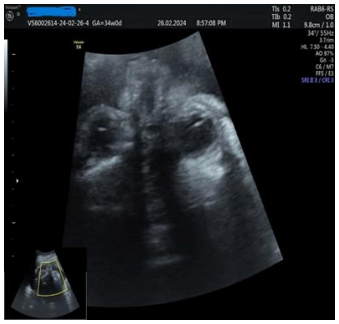
Picture 5: 45, XO It was confirmed with the microarray.



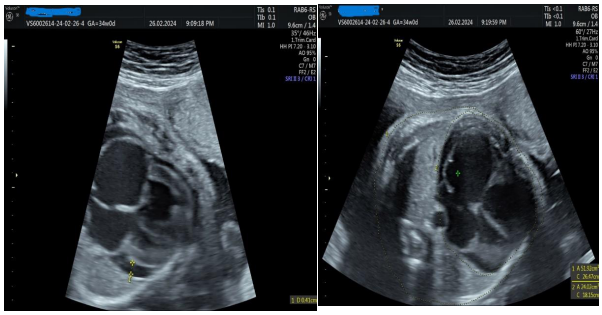
Picture 6: Color Doppler of the fetal head.



Picture 7: Fetal ear.



Picture 8: Fetal face (perpendicular plane).



Picture 9: Fetal heart (cardiomegaly).

References

1. Pooh RK, Degani S (2012) Fetal cerebral circulation. In: Timor-Tritsch IE,Monteagudo A, Pilu G, Malinger G, eds. Ultrasonography of the prenatal brain, 3rd ed. New York, NY: The McGraw-Hill Company 428-446. Link: https://bit.ly/3YV7jMX

2. Recinos PF, Rahmathulla G, Pearl M, Violette Renard R, George IJ, et al. (2012) Vain of Galen malformations: epidemiology, clinical presentations, management. Neurosurg Clin N Am 23: 165-177. Link: https://bit.ly/3UY1Q71

3. Ali Abdelghany WM, Nouby R, Ramadan OM, Habib MA (2024) Endovascular treatment of vein of Galen aneurysmal malformation: hospital-based case series in two tertiary centers. Egypt J Neurol Psychiatry Neurosurg 60: 49. Link: https://bit.ly/3UYzw4o

4. Makishima T, King K, Brewer CC, Zalewski CK, Butman J, et al. (2009) Otolaryngologic markers for the early diagnosis of Turner syndrome. Int. J. Pediatr. Otorhinolaryngol 73: 1564-1567. Link: https://bit.ly/40XDoGv

5. Gravholt CH, Juul S, Naeraa RW, Hansen J (1998) Morbidity in Turner syndrome. J. Clin. Epidemiol 51: 147-158. Link: https://bit.ly/4ftyWnk

6. Elsheikh M, Dunger DB, Conway GS, Wass JA (2002) Turner's syndrome in adulthood. Endocr. Rev 23: 120-140. Link: https://bit.ly/3UZOyXx