Cerebral Bleed in Turner’s Syndrome: A Rare Presentation

Dr. Urvee Swaika, Dr. Dhruti Amrutiya, Dr. Priyanka Amonkar, Dr. Jeetendra Gavhane

Abstract: We report an unusual case of a 13 year old female with CT Brain findings of intraventricular and thalamic bleed, with short stature, diagnosed as Turner syndrome on karyotyping (mosaic pattern). For decades, repeated epidemiologic observations have been made regarding the inverse relationship between stature and cardiovascular disease, including stroke. Some cases of cerebral hemorrhage have been reported among patients with Turner syndrome. In most of these cases, it resulted from renovascular hypertension. Notably, our patient did not have hypertension, still developed a bleed. Very few cases of intracranial hemorrhage without hypertension have been reported, with the probable cause being a connective tissue defect.

Keywords: Short stature, cerebral bleed, Turner’s syndrome

1. Case Presentation

A 13 year old female had come with complaints of severe headache since 4 days, more in the frontal region; 3 episodes of non projectile vomiting, one undocumented fever spike. Child did not have history of any convulsion episode/bleeding/neurological deficit. On examination, child was irritable with a GCS of 13/15 (E4M5V4) with a subtle left upper limb motor weakness without any hypertension or meningeal signs. CT Brain was done suggestive of small intraventricular bleed in 3rd ventricle and adjacent right thalamic region measuring 1.5*1.3 cm extending into both lateral ventricles with a normal PT INR APTT. On further detailed history and examination, she was found to have short stature, with mild delay in puberty. Karyotyping was sent which had 34 metaphases showing monosomy of X chromosome and 16 showing 46 XX pattern, overall suggestive of mosaic pattern of Turner’s syndrome.

Figure 1: CT Brain of the patient showing small intraventricular bleed in 3rd ventricle and adjacent right thalamic region measuring 1.5*1.3 cm extending into both lateral ventricles

Figure 2: Clinical photo of the patient

2. Discussion

Turner’s syndrome is a chromosomal disorder caused by complete deletion of an X chromosome or a mosaic pattern. Signs commonly noted are weblike neck, low set ears, broad chest with widely spaced nipples, wide carrying angle, short 4th metacarpal, streak ovaries. On the short arm of the X chromosome, the pseudoautosomal region (PAR-1) is involved and the short stature homeobox (SHOX) gene is located in this region. This gene deficiency is known to contribute to the short stature or skeletal deformities of Turner patients.

Several complications such as congenital heart disease, hypothyroidism, strabismus, sensorineural hearing loss, renal malformations have been noted. The most common heart defect is bicuspid aortic valve. Hypertension is also a common complication and associated vascular complications like aortic dissection/ cerebral hemorrhage have been studied. In all cases, the patients either have XO or mosaic type with only one study showing deletion of short arm of X chromosome. (2)

Some cases of ischemic stroke and less commonly, embolic stroke have also been reported in Turner’s syndrome patients.

The most common causes for cerebral hemorrhage are hypertension, a clotting defect, or a general connective tissue defect. With regards to hypertension, PRA (plasma renin activity) is higher in Turner’s compared to controls. This may be due to renal vascular abnormality. (3) One case of a 51 year old with Turner’s (non mosaic pattern) has been published as a case of spontaneous cerebral hemorrhage without hypertension. (1) Here, the patient had hyper flexible joints and hyper extensible skin and ectatic ascending aorta and brachiocephalic trunk on

Volume 12 Issue 1, January 2023

www.ijsr.net
Licensed Under Creative Commons Attribution CC BY

Paper ID: SR23105163711
DOI: 10.21275/SR23105163711
229
angiography, a general connective tissue defect was suspected, making arteries more prone to physiologically increased blood pressure and rupture of intracerebral arteries with consecutive bleeding.

In another case, recombinant human GH (rhGH) therapy was initiated from the age of 11 years and completed at 15 years. Following the rhGH therapy, female sex steroid replacement therapy was started. However, sex hormone replacement therapy is not recognised as elevating the blood pressure.

Nathwani et al (4) had reported that most patients with Turner’s have an abnormal blood pressure circadian rhythm, suggesting that it is relatively difficult to detect the presence of hypertension in patients with Turner’s.

Fudge et al (5) in contrast, reported the significance of ambulatory blood pressure monitoring for detecting hypertension in such a population.

3. Conclusion

Patients with Turner’s syndrome should carefully monitor their blood pressure and avoid emotional stress, competitive sports and sports associated with risk of trauma. The possibility of Turner’s syndrome should be considered in all cases where there has been a lethal cerebrovascular event in younger women.

References