

Severity of Ventriculomegaly and its Associated Finding using Antenatal Ultrasound Scan at a Tertiary Care Hospital: A Retrospective Descriptive Study

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ABSTRACT

Introduction: Foetal Ventriculomegaly (VM) is defined as the enlargement of the lateral ventricles in the developing foetal brain. The measurement of foetal cerebral lateral ventricles' size is routinely recommended as part of the second-trimester foetal scan to screen for anomalies. VM can occur due to obstruction of the Cerebrospinal Fluid (CSF) tract, absorption disorders, inadequate brain development, or excessive CSF production.

Aim: To classify VM based on severity to differentiate between isolated and non isolated VM.

Materials and Methods: A retrospective descriptive study was conducted in the Department of Radiology at PESIMSR, Andhra Pradesh, India., a tertiary medical hospital, from April 2018 to April 2021. Cases were selected from computer records showing a diagnosis of hydrocephalus or foetal VM in patients who underwent the Ultrasound (US) anomaly scan. VM was defined as a lateral ventricle width of ≥ 10 mm. Foetal VM was classified, and associated CNS and extracranial abnormalities and their details were collected. Descriptive statistics were used, and results were expressed in terms of frequency and percentage.

Results: The VM was observed in 91 fetuses, with mild VM in 59 fetuses (64%), moderate VM in 14 fetuses (15.2%), and severe VM in 18 fetuses (19.5%). The median age at diagnosis was 26.5 weeks. Isolated mild VM was seen in 26 cases (44%), and it was associated with other abnormalities in 33 cases (56%). Isolated moderate VM was observed in four cases (28.5%), while 10 cases (71.4%) were associated with other abnormalities. Isolated severe VM was seen in (33.3%) 6, and it was associated with abnormalities in 12 cases (66.6%). The most common associated abnormalities were foetal growth restriction (15 cases), absent cavum septum pellucidum (eight cases), and open spina bifida, Arnold-Chiari malformation (eight cases).

Conclusion: Ultrasound is a sensitive tool for detecting VM. Once VM is diagnosed, a detailed examination of the fetus should be conducted to evaluate other CNS and non CNS anomalies, thereby differentiating isolated from non-isolated VM, as the prognosis may vary. This information will be valuable for counseling patients.

Keywords: Arnold-Chiari malformation, Cavum septum, Isolated anomaly, Spina bifida

INTRODUCTION

Lateral Ventriculomegaly (VM) is the most common abnormality of the foetal nervous system. Measurement of the size of the foetal cerebral lateral ventricles is recommended as part of the routine foetal scan performed during the second trimester to screen for foetal anomalies [1]. VM can be caused by obstruction of the CSF tract, absorption disorders, inadequate brain development, and excessive buildup of CSF. However, managing the condition and counseling parents is still difficult because the cause, absolute risk, and degree of resulting neurological deficit cannot be determined with confidence [2].

The prevalence of VM ranges from 0.3 to 1.5 per 1,000 births in different series. VM is more common in males, with a male-to-female sex ratio of 1.7 [2,3].

The largest multicentre study in Europe (EUROFETUS) [4] reported a sensitivity of 93.5% for diagnosing foetal VM by ultrasound. Regardless of the degree of VM (mild or severe), it is important to conduct a thorough search for Central Nervous System (CNS) and extra CNS anomalies. Evaluation of the foetal karyotype is recommended, and a search for foetal infections should also be performed. Even in the presence of a normal karyotype, absence of infection, and apparently normal foetal anatomy, when counseling the parents, one should be aware of the limited accuracy of ultrasound in distinguishing isolated from non isolated VM [3].

Furthermore, the possibility of VM progression and late onset brain anomalies should be taken into account. Hence serial ultrasound should be planned and need for fetal MRI should be considered. Even in cases with apparently normal findings, parents must be informed about the possibility that some anomalies or syndromes cannot be diagnosed before birth [3]. Hence, the present study was conducted to classify VM based on severity and differentiate isolated from non isolated VM.

MATERIALS AND METHODS

A retrospective descriptive study was conducted in the Department of Radiology at PES Institute of Medical Sciences and Research, Kuppam, Andhra Pradesh, India, between April 2018 and April 2021. Cases were selected from computer records showing a diagnosis of either hydrocephalus or foetal VM.

Inclusion criteria: The data of fetuses, both in house delivered and referral cases, who came for anomaly scans in the centre and were diagnosed with VM (at least one lateral ventricle with a width of ≥ 10 mm), were included [5].

Exclusion criteria: Cases diagnosed with foetal cytomegalovirus or toxoplasma infection and those with abnormal genetic tests were excluded from the study.

Study Procedure

A total of 91 foetal cases were collected for the study. Foetal VM was classified as follows:

- Mild: lateral ventricle measurement of 10-11.9 mm.
- Moderate: lateral ventricle measurement of 12-14.9 mm.
- Severe: lateral ventricle measurement of ≥ 15 mm [5].

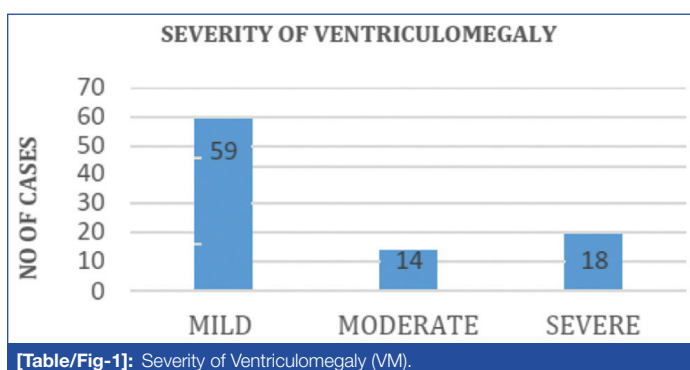
Data on CNS and extracranial abnormalities and their details were also collected [5].

STATISTICAL ANALYSIS

The data was entered into Microsoft Excel, and descriptive statistics were used, including frequency (n) and percentages.

RESULTS

A retrospective review of VM reports was conducted on 91 cases from the study centre's database. Among these cases, there were 59 cases of mild VM, 14 cases of severe VM, and 18 cases of moderate VM [Table/Fig-1]. Most of the cases (68.4%) showed dilatation of both lateral ventricles, while 30% of cases exhibited dilatation in only one ventricle. In the unilateral cases, the left-side ventricle was predominantly affected (67.85%), followed by the right-side ventricle (32.14%).



[Table/Fig-1]: Severity of Ventriculomegaly (VM).

Out of the study population, 55 cases (60%) fell within the age group of 21-29 years, and 26 cases (24%) were in the age group of 30-35 years. Two cases (2.19%) involved twins with VM during the study period, and 89 cases (98%) were singleton pregnancies.

Isolated lateral ventricle involvement was observed in 55 cases (60.44%), while 36 cases (39.56%) exhibited other associated abnormalities, highlighting the need for a detailed evaluation of the foetus to rule out cranial and extracranial abnormalities.

Among the mild VM cases, which accounted for 64.2% of the study population (59 cases), 33 cases (56%) showed unilateral VM, and 26 cases (44%) exhibited bilateral VM. Isolated VM was observed in 26 cases (44%), while 33 cases (56%) had VM associated with other abnormalities. The associated abnormalities are listed in [Table/Fig-2], with craniofacial and skeletal abnormalities being the most common structural abnormalities noted.

System involved	Associated malformations	n	Percentage (%)
CNS	Absent cavum septum pellucidum	2	3.38
	Small cavum septum pellucidum	1	1.69
	Corpus callosum agenesis	2	3.38
	Persistent Blake's pouch cyst	2	3.38
	Possibly evolving lissencephaly	2	3.38
	Colpocephaly	1	1.69
	Strawberry-shaped head	1	1.69
	Inferior vermian hypoplasia	1	1.69
	Mega cisterna magna	1	1.69
	Microcephaly	1	1.69
	Multiple discrete hyperechoic foci seen in bilateral frontal and parietal cortices	1	1.69

Face	Micrognathia	4	6.77
	Bilateral left lip and palate	2	3.38
	Flat facies	2	3.38
	Non ossified nasal bone	1	1.69
Thorax	Congenital high airway obstruction syndrome	1	1.69
	Diaphragmatic hernia	1	1.69
Cardiac	Axis deviation	2	3.38
	Mild global cardiomegaly	1	1.69
	Ventricular septal defect	6	10.16
	Echogenic intracardiac focus	8	13.56
	Persistent left Superior Vena Cava (SVC)	1	1.69
	Coarctation of aorta	1	1.69
GIT	Oesophageal atresia/fistula	2	3.38
Gut	Renal pelvic dilatation	4	6.78
	Horse-shoe kidney	1	1.69
Musculoskeletal system	Bilateral Congenital Talipes Equinovarus (CTEV)	3	5.08
	Unilateral club foot	2	3.38
	Flexed abnormality of wrist	1	1.69
	Polydactyl	2	3.39
	Saddle gap in the right foot	1	1.69
	Hyperextension at tarsometatarsal joint of the third toe in the right foot	1	1.69
Spine	Spina bifida meningomyelocele	4	6.77
Doppler	Umbilical artery abnormal	2	3.39
	Middle cerebral artery abnormality	2	3.39
	Tricuspid regurgitation	1	1.69
	Uterine artery doppler abnormal	4	6.77
Growth	Foetal growth restriction	8	13.56
Liquor	Oligohydramnios	1	1.69
	Polyhydramnios	3	5.08
	Anhydramnios	1	1.69
SUA	Single Umbilical Artery	4	6.77
Others	Increased nuchal fold thickness	4	6.77
	Amniotic band	2	3.39
	Massive foetal ascites	1	1.69

[Table/Fig-2]: Associated malformations in mild Ventriculomegaly (VM) (n=59).

There were 14 cases of moderate VM, constituting 15.2% of the study population. Among these cases, 2 cases (14.2%) showed unilateral VM, and 12 cases (85.7%) exhibited bilateral VM. Isolated VM was observed in 4 cases (28.5%), while 10 cases (71.4%) had VM associated with other abnormalities. The associated abnormalities are presented in [Table/Fig-3].

There were 18 cases of severe VM, constituting 19.5% of the study population. All of these fetuses exhibited bilateral VM. Isolated VM was observed in 6 cases (33.3%), while 12 cases (66.6%) had VM associated with other abnormalities [Table/Fig-4].

System involved	Associated malformations	n	Percentage (%)	
CNS	Large anterior cerebral arterial aneurysm	1	7.14	
	Bilateral choroid plexus cyst	2	14.28	
	Lemon-shaped skull	1	7.14	
	Partial agenesis of corpus callosum	1	7.14	
	Agenesis of corpus callosum	1	7.14	
	Evolving lissencephaly	1	7.14	
	Absent cavum septum pellucidum	2	14.28	
	Corpus callosal dysgenesis	2	14.28	
	Cardiac	Global cardiomegaly	1	7.14
		Echogenic intracardiac foci	2	14.28

Gut	Renal pelvic dilatation	1	7.14
Spine	Lumbar kyphoscoliosis	1	7.14
	Arnold-Chiari malformation type II	1	7.14
	Lumbar open spina bifida-extending from L2 to L5	1	7.14
Doppler	Umbilical artery abnormal	2	14.28
	Middle cerebral artery abnormal	1	7.1
	Uterine artery Doppler abnormality	1	7.14
Growth	Foetal growth restriction	4	28.57

[Table/Fig-3]: Malformations in moderate Ventriculomegaly (VM) (n=14).

System involved	Associated malformations	n	Percentage (%)
CNS	Absent cavum septum pellucidum	3	16.66
	Partial agenesis of corpus callosum	2	11.11
	Absent corpus callosum	1	5.55
	Hypoplastic cerebellum	2	11.11
	Complete vermian agenesis	1	5.55
	Strawberry-shaped head	1	5.55
	Lemon-shaped head	1	5.55
	Persistent Blake pouch cyst	1	5.55
	Choroid plexus cyst	1	5.55
	Mega cisterna magna	2	11.11
Face	Hypoplastic nasal bone	1	5.55
	Depressed nasal bridge	1	5.55
	Flat facies	1	5.55
Cardiac	Perimembranous VSD	2	11.11
	Tricuspid dysplasia	1	5.55
	Hypoplastic right ventricle	1	5.55
	Small pulmonary artery	1	5.55
Gut	Unilateral dysplastic kidney	1	5.55
	Renal pelvic dilatation	1	5.55
Spine	Open spina bifida with Arnold-Chiari II malformation	3	16.66
Musculoskeletal system	Bilateral rocker bottom feet	1	5.55
Doppler	Uterine artery doppler abnormality	1	5.55
	Umbilical artery doppler abnormality	1	5.55
	MCA doppler abnormality	1	5.55
Growth	Foetal growth abnormalities	3	16.66
Liquor	Polyhydramnios	2	11.11
SUA	Single Umbilical Artery	2	11.11
Others	Increased nuchal fold thickness	1	5.55

[Table/Fig-4]: Malformations in severe Ventriculomegaly (VM) patients (n=18).

VSD: Ventricular septal defect

DISCUSSION

In present study, the authors retrospectively analysed ultrasound scans of fetuses diagnosed with foetal VM over a period of three years. The majority of cases in the study were mild VM, with unilateral mild VM being more common. Non-isolated mild VM was more common than isolated mild VM. Therefore, a detailed foetal evaluation is necessary to determine if it is isolated VM, considering the diagnostic factors mentioned in previous studies [6,7]. Severe VM was associated with 100% bilateral dilatation and can result in serious outcomes [8]. It was observed that CNS findings were more common in fetuses with symmetric VM, similar to the study by Morris JE et al., [6].

The present study is consistent with previous studies that found more CNS anomalies in moderate and severe VM compared to mild VM [9,10].

Foetal MRI is an excellent diagnostic modality that enables the detection of additional abnormalities in approximately 50% of VM cases, such as agenesis of the corpus callosum, absence of the septum pellucidum, disorders of cortical development, and cerebrovascular abnormalities [7]. It is more sensitive than ultrasonography for detecting cortical malformation, haemorrhage, and parenchymal disorders. However, not all of these findings may be clinically significant or associated with a poor prognosis, but early identification is helpful for parental counseling and further follow-up [6].

The VM may be associated with foetal infections in approximately 2% of cases, particularly Cytomegalovirus (CMV), toxoplasmosis, parvovirus, and Zika virus [7]. Many cases of VM due to infectious diseases present with other sonographic findings, such as brain calcifications, periventricular cysts, or extracranial signs. However, in some cases, VM is the only sonographic feature [5]; therefore, testing for infections should be offered in all cases of isolated VM and strongly recommended if the foetus has other features suspicious for an infectious cause [6]. Maternal serology is the first diagnostic test, but it is less sensitive and specific than Polymerase Chain Reaction (PCR) on amniotic fluid. Thus, amniotic fluid PCR is the preferred method of evaluation for foetal infections [6,8].

Limitation(s)

The retrospective nature of the study is an inherent limitation. Data on neurodevelopmental outcomes were not assessed.

CONCLUSION(S)

Ultrasound is a sensitive tool for detecting VM. A systematic and detailed study of the foetus during the anomaly scan is essential. Once VM is diagnosed, a detailed examination of the foetus should be performed to evaluate other CNS and non-CNS anomalies. Differentiation between isolated and non isolated VM is crucial as the prognosis varies, and it helps in counseling the patients. This study emphasises the importance of proper patient follow-up. However, since the presence of CNS abnormalities has been shown to be closely related to the neuro developmental outcome of the child, findings in foetal ultrasound imaging can be used as an indication of the outcome in cases where follow-up is lost.

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