

## Association of fetal cerebral ventriculomegaly (VM) with chromosomal and structural anomalies - Does laterality and size matter?

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### Objective

Ventriculomegaly is an easily identifiable ultrasound finding in the second trimester anomaly scan. The etiology of fetal VM is diverse and prognosis of the pregnancy is variable. The aim of study is to evaluate fetuses with cerebral ventricular size of more than or equal to 10mm for the presence of chromosomal and/or genetic anomalies and associated intra and extra cranial anomalies.

### Methods

This is a retrospective study of prospectively collected data conducted at a tertiary fetal care centre in South India. The study period was from January 2010 till December 2021. All singleton pregnancies with fetal cerebral ventricle measurement more than or equal to 10mm (ventriculomegaly, VM) at 18<sup>+0</sup> – 24<sup>+0</sup> weeks' scan were evaluated further for the presence of chromosomal, other intra and extra cranial anomalies. The cerebral ventricles were measured in the axial view of the transventricular plane at the level of parieto-occipital fissure with calliper placement from inner to the inner border. The proximal ventricle was also measured in all the fetuses as per the unit protocol. All fetuses with VM had a further detailed assessment for presence of intracranial (neurosonogram) and extra cranial anomalies. Parents were offered invasive testing for fetal chromosomal and/ or genetic testing as was indicated. The data was analyzed on chi-square test to assess the significance. Ultrasound scans were performed by FMF certified operators for the 18 – 24 weeks' scan and examinations were recorded on the Astraia fetal database software. Outcome of the pregnancy was obtained by telephonic conversation with the parents and examination of the delivery details in the hospital records.

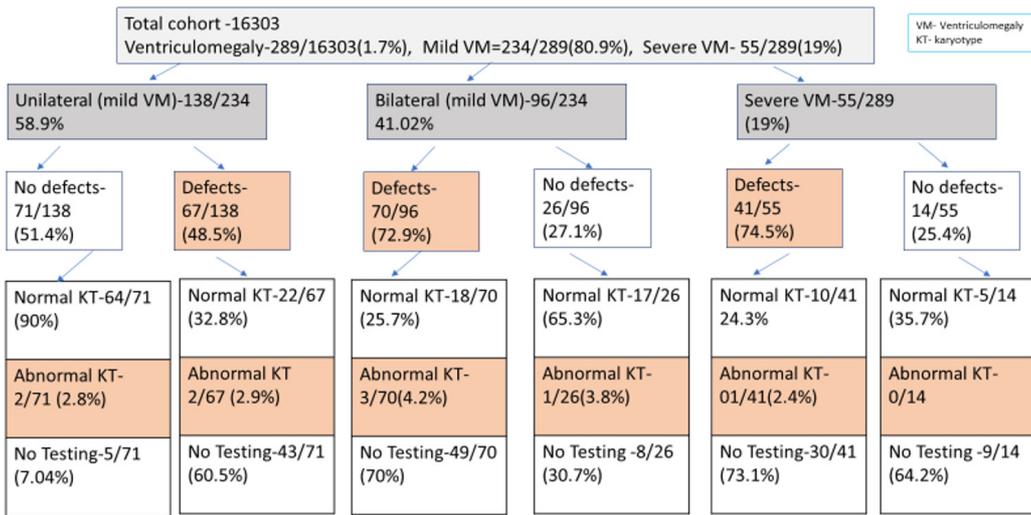
### Results

289/16,303 (1.7%) fetuses had ventriculomegaly. 234/ 289 (80.9%) fetuses had mild VM (10 – 15mm), of which 138/ 234 (58.9%) was unilateral and 96/ 234 (41.1%) was bilateral MVM. 32/138 (23.2%) and 22/96 (22.9%) had other intracranial defects in the unilateral and bilateral groups respectively. 35/138 (25.3%) and 48/96 (50%) in the unilateral and bilateral groups had associated anomalies in other systems respectively. (p value=.078, NS). Fetal chromosomal analyses was available for 129/234 (55.1%) fetuses. 4/90 (4.4%) and 4/39 (10.2%) had a chromosomal abnormality in the unilateral (2 x T21, 1 x partial Trisomy 8 with monosomy 2, 1 x GPSM 2 mutation) and bilateral (2 x T21, 1x skeletal dysplasia gene mutation, 1 X Bardet Biedl syndrome) groups respectively. Of these 8 fetuses, 3 were isolated MVM, all 3 had trisomy 21, giving an incidence of 4.7% (3/ 64). 55/ 289 (19.1%) fetuses had severe VM (> /= 16mm), ie. unilateral, bilateral or tri VM. 15/55 (27.2%) had other intracranial defects. 26/55 (47.2%) had associated anomalies in other systems. Fetal chromosomal and / or genetic analysis was available for 16/55 (29.1%) fetuses, of which 1/ 16 (6.2%) had a genetic abnormality (GPSM2 mutation gene) and this was isolated SVM.

### Conclusion

Fetal cerebral ventriculomegaly is an easily identifiable anomaly at the 18 – 24 weeks' scan. The outcome of the pregnancy depends on the size, laterality of the ventricles, and the presence or absence of associated chromosomal and non-chromosomal anomalies. In our study, we have confirmed that unilateral and bilateral mild ventriculomegaly can be associated with chromosomal and non-chromosomal anomalies. Hence, on detection of fetal cerebral ventricular size of more than or equal to 10mm as measured in the axial plane of the fetal head, it is paramount to consider possibility of fetal chromosomal anomalies even when isolated and or unilateral. In our study, 3/ 9 (33.3%) fetuses with abnormal karyotype had isolated MVM. The presence of associated anomalies will further worsen the outcome for these fetuses. The incidence of associated anomalies was higher in the severe and bilateral mild groups (p value =0.002 significant). However, even unilateral mild VM in euploid fetuses was associated with intra and extra cranial defects in about 25.5% in our study. We conclude that, VM is an important precedent to investigate the fetus. All pregnancies with fetal VM must be assessed in detail for the presence of associated intra and extra cranial anomalies and in addition, offered prenatal invasive testing for the presence of chromosomal and or genetic anomalies, even when isolated or unilateral. The strength of our study is that we have categorized VM according to the severity and laterality to assess the significance of association with chromosomal /genetic and structural abnormalities. The limitation of the study is exclusion of fetuses which underwent termination without evaluation for chromosomal or genetic anomalies.

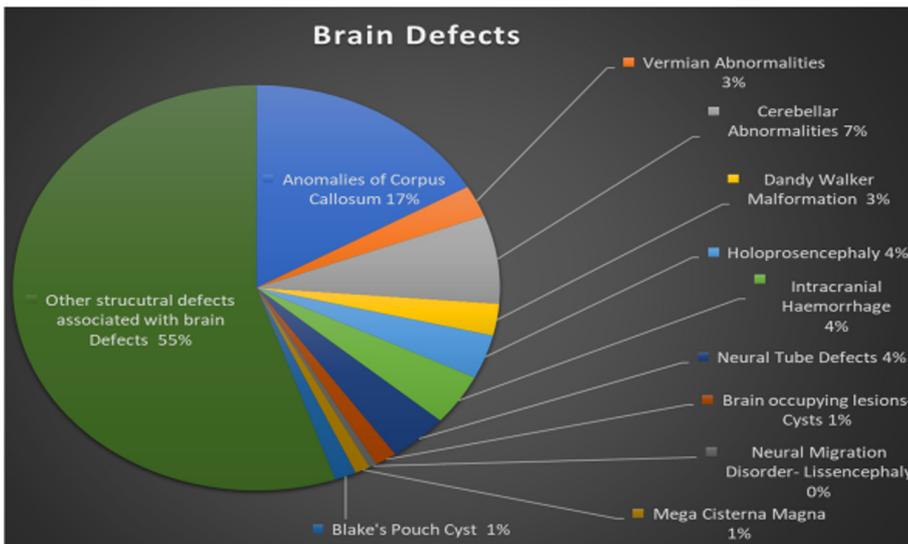
( ID:4082) Table 1- Associated structural and chromosomal abnormalities in fetuses with Ventriculomegaly



( ID:4082) Table 2-Outcome of fetuses with Ventriculomegaly

Total cohort -16303 Ventriculomegaly (VM)- 289/16303(1.7%) VM with known KT=145/289(50.1%)	ABNORMAL KARYOTYPE (KT)	Normal KT	Structural defects in Euploid fetuses	No Defects with Euploid fetuses	Outcome
Unilateral VM (mild) 90/138(65.2%)	4/90 (4.4%)	86/90 (95%)	22/86 (25.5%)	64/86 (74.4%)	Total live birth-63/64 (98.4%) Intrauterine Death(IUD) -1/64 (1.56%)
Bilateral VM (mild) 39/96(40.6%)	4/39 (10.2%)	35/39 (89.7%)	18/35 (51.4%)	17/35 (48.5%)	Termination -2/17(6%) Live birth-13/15(86.6%) Neonatal Death(NND)-1/15(6.6%) IUD-1/15(6.6%)
Severe VM 16/55(29%)	1/16 (6.2%)	15/16 (93.7%)	10/15 (66.6%)	5/15 (33.3%)	Termination-2/5(40%) Live birth -2/3(66.6%) NND-1/3(33.3%)

( ID:4082) Fig 1 -Brain defects in fetuses with Ventriculomegaly



( ID:4082) -Images showing cerebral ventriculomegaly

