Pakistan Journal of Medical & Cardiological Review
https://pakjmcr.com/index.php/1/about
Online ISSN 3007-2387
Print ISSN 3007-2379

Prenatal Diagnosis and Pregnancy Outcomes in Fetuses with Ventriculomegaly: A Systematic Review

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Keywords:

Received on 01 May 2026
Accepted on 22 May 2026
Published on 11 Jun 2026

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Abstract

Fetal ventriculomegaly (VM) is one of the most common prenatal central nervous system abnormalities detected during obstetric ultrasonography. Prognosis varies depending on ventricular size, associated anomalies, chromosomal abnormalities, and progression during pregnancy. Recent advances in fetal MRI and chromosomal microarray analysis (CMA) [1,5] have improved diagnostic accuracy and prenatal counseling.

Introduction

Fetal ventriculomegaly (VM) is one of the most frequently diagnosed abnormalities of the fetal central nervous system (CNS) detected during prenatal ultrasonography. It is defined as dilatation of the lateral cerebral ventricles measuring 10 mm or greater at the level of the atrium and is identified in approximately 0.3–2 per 1,000 pregnancies [1,2]. As one of the most common prenatal CNS abnormalities, ventriculomegaly represents a major indication for referral to fetal medicine specialists and advanced prenatal neuroimaging. The condition may occur as an isolated finding or in association with other CNS malformations, congenital infections, chromosomal abnormalities, and various genetic or syndromic disorders, making its diagnosis and prognosis highly variable.

Ventriculomegaly is commonly classified according to ventricular width into mild (10–12 mm), moderate (13–15 mm), and severe (>15 mm) forms [3]. This classification has important prognostic implications, as the risk of neurological morbidity, developmental impairment, and perinatal mortality generally increases with the degree of ventricular enlargement. Approximately 60–70% of prenatally diagnosed cases are classified as mild ventriculomegaly, which is usually associated with favorable neurodevelopmental outcomes when isolated [4]. Nevertheless, evidence suggests that 10–15% of apparently isolated mild cases may subsequently develop subtle cognitive, behavioral, or neurodevelopmental abnormalities during childhood. In contrast, severe ventriculomegaly is frequently associated with

hydrocephalus, cortical thinning, seizures, developmental delay, and increased perinatal mortality [5].

The etiology of fetal ventriculomegaly is highly heterogeneous. Common underlying causes include structural CNS abnormalities, congenital infections such as cytomegalovirus and toxoplasmosis, intracranial hemorrhage, and chromosomal abnormalities. Associated anomalies are reported in approximately 50–70% of non-isolated cases, significantly influencing prognosis and management. Chromosomal abnormalities are identified in nearly 5–20% of fetuses with ventriculomegaly, with trisomy 21, trisomy 18, and pathogenic copy number variants among the most frequently reported genetic findings. Recent studies have demonstrated that chromosomal microarray analysis (CMA) provides an additional diagnostic yield of approximately 8–12% beyond that achieved with conventional karyotyping alone, highlighting the increasing importance of advanced genetic testing in prenatal evaluation [1,5].

Advances in prenatal imaging have substantially improved the diagnostic assessment of fetal ventriculomegaly. Although ultrasonography remains the primary diagnostic modality, fetal magnetic resonance imaging (MRI) has emerged as a valuable complementary tool for identifying additional CNS abnormalities that may not be detected on ultrasound examination [2,3]. Studies have shown that MRI can reveal associated abnormalities in approximately 5–20% of cases initially considered isolated on prenatal sonography. Furthermore, serial prenatal imaging has demonstrated progression of ventricular dilatation in approximately 13–16% of affected fetuses, a finding that is strongly associated with adverse neurological and developmental outcomes.

Pregnancy outcomes in fetuses with ventriculomegaly vary considerably and are influenced by several factors, including ventricular size, progression of ventricular dilatation, laterality, associated structural anomalies, and underlying genetic abnormalities. Survival rates in cases of isolated mild ventriculomegaly exceed 90%, whereas severe ventriculomegaly may be associated with mortality rates ranging from 20% to 50%, particularly when accompanied by additional CNS abnormalities [4,6]. Consequently, accurate prenatal diagnosis, comprehensive genetic evaluation, and multidisciplinary assessment are essential for parental counseling, clinical decision-making, pregnancy management, and prediction of long-term neurodevelopmental outcomes.

Given the substantial heterogeneity of fetal ventriculomegaly and the recent advances in prenatal imaging and genomic technologies, a comprehensive synthesis of the available evidence is warranted. Therefore, this systematic review aims to evaluate current evidence regarding the prenatal diagnosis, genetic evaluation, pregnancy outcomes, and long-term prognosis of fetuses diagnosed with ventriculomegaly.

Methods

This systematic review was conducted according to PRISMA 2020 guidelines. Electronic databases including PubMed, Scopus, Web of Science, SpringerLink, and Google Scholar were searched for studies published after 2023.

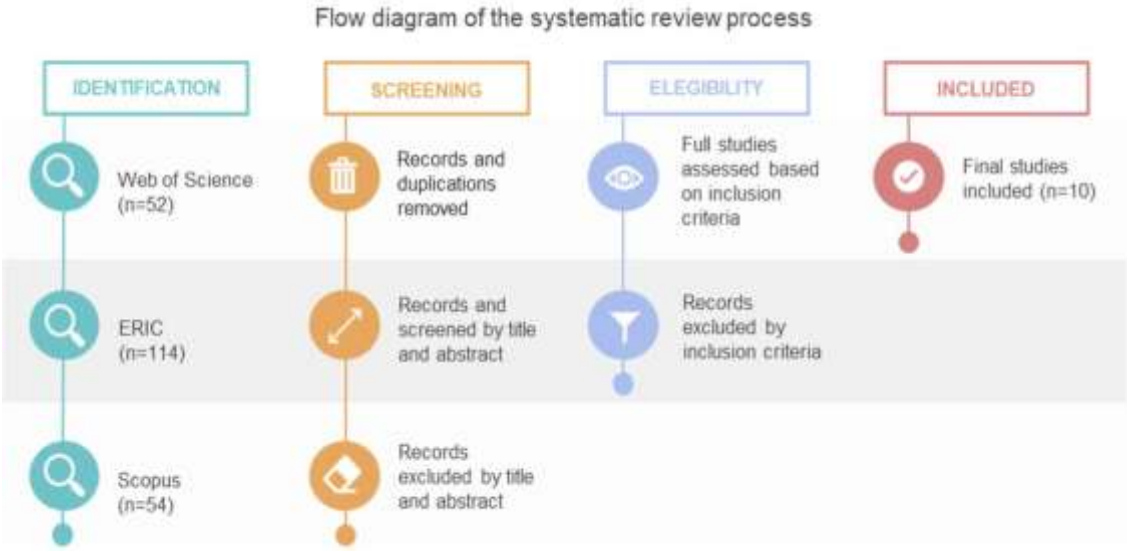
Inclusion Criteria

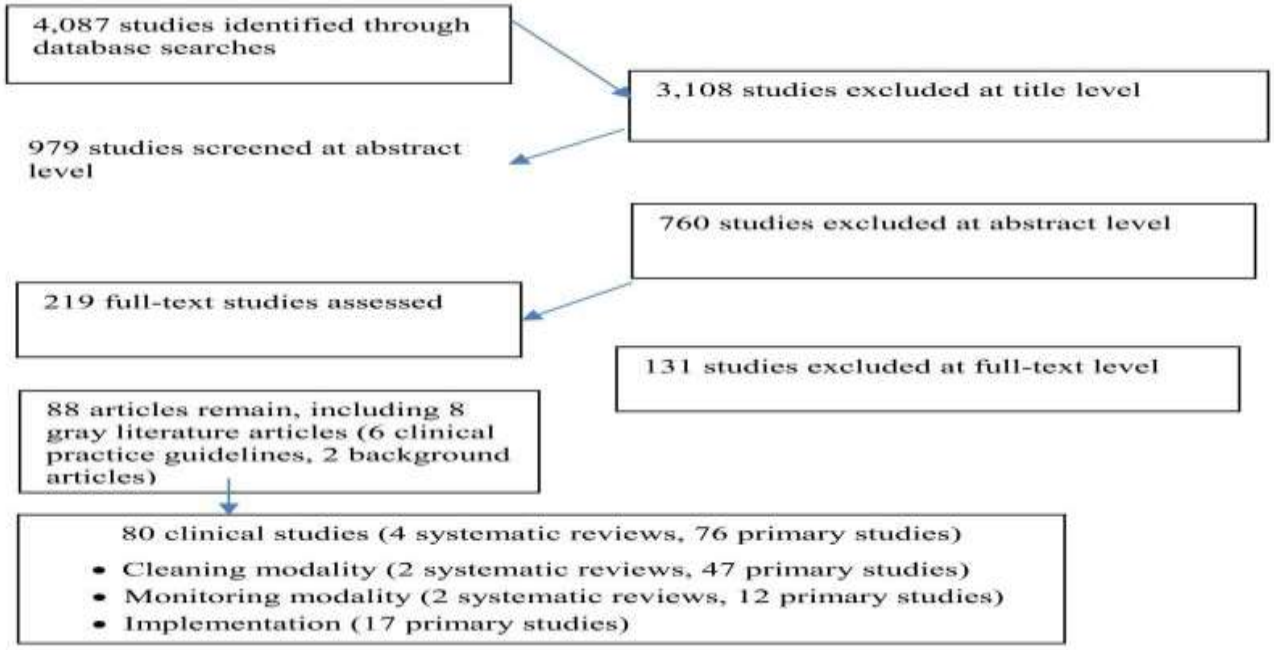
Prenatally diagnosed ventriculomegaly
English-language studies
Cohort studies, systematic reviews, and meta-analyses
Studies reporting pregnancy or neurodevelopmental outcomes

Exclusion Criteria

Case reports
Animal studies

Duplicate publications
Studies lacking outcome data





Prenatal Ultrasound Diagnosis

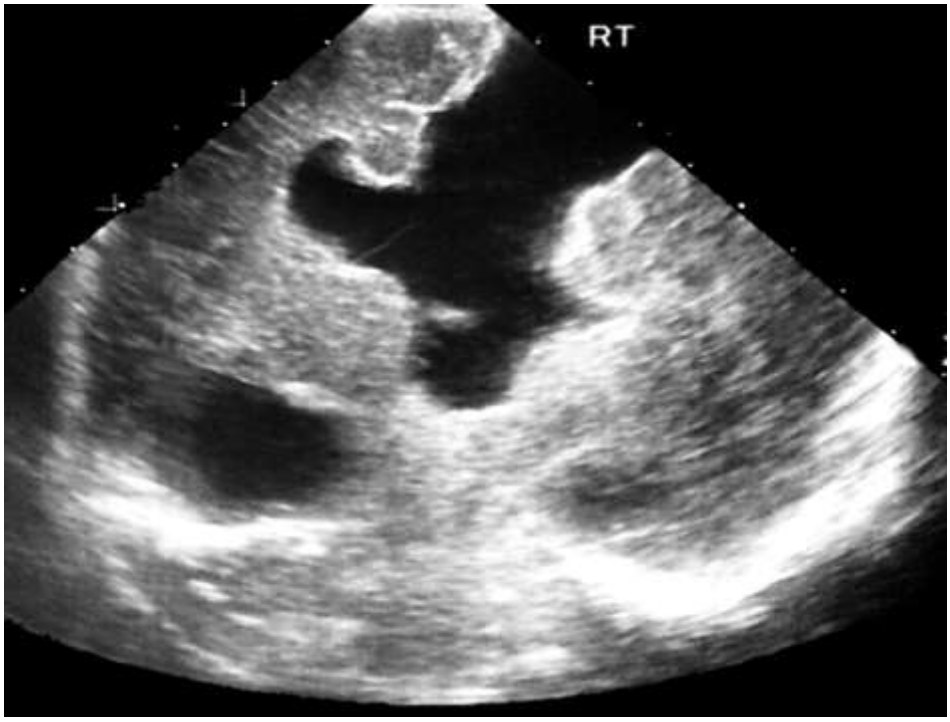
Ultrasound remains the primary modality [1,2] for diagnosis of fetal ventriculomegaly. Measurements are obtained at the atrium of the lateral ventricle in the axial transventricular plane.

Common sonographic findings include:

Enlarged lateral ventricles
Dangling choroid plexus
Bilateral ventricular enlargement
Midline abnormalities
Posterior fossa anomalies

Mild Ventriculomegaly

Mild ventriculomegaly (10–12 mm) [3,4] is frequently associated with favorable neurodevelopmental outcomes [4], especially when isolated.



Severe Ventriculomegaly and Hydrocephalus
Severe ventriculomegaly (>15 mm) [5,6] is associated with:
Hydrocephalus
Cortical thinning
Developmental delay
Increased perinatal mortality
Progressive ventriculomegaly and bilateral involvement are important predictors of poor outcome.

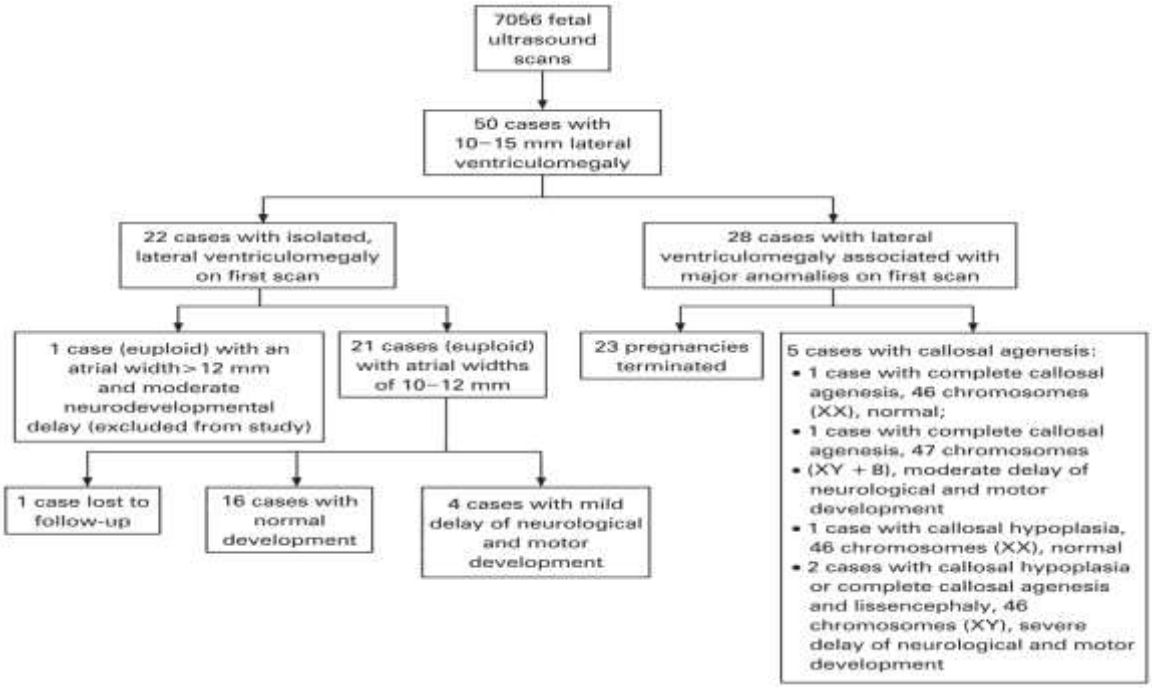


TABLE 5. Clinical details of the 12 cases with developmental delay

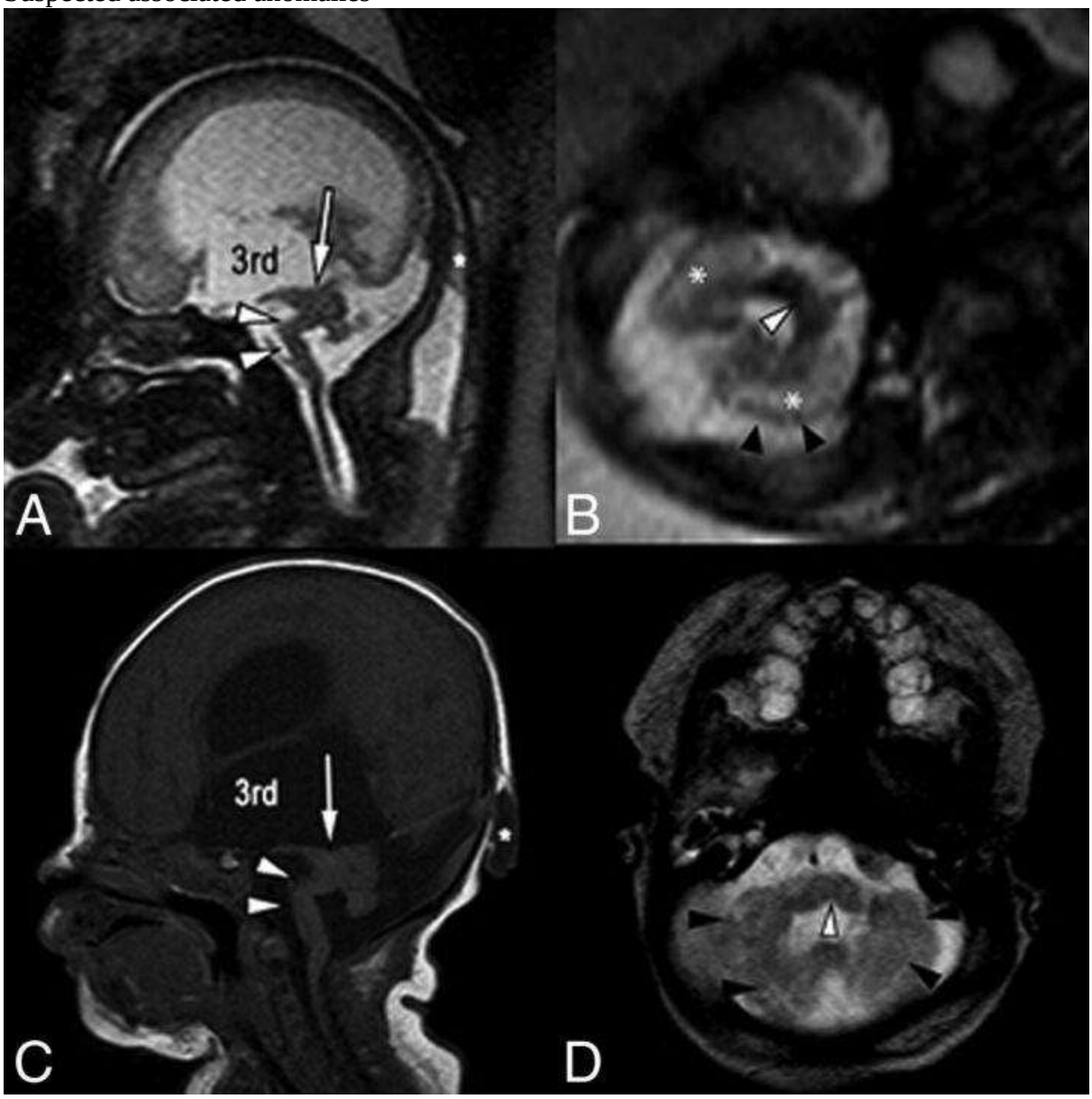
Case	Degree of fetal ventriculomegaly	Prenatal structural abnormalities	Karyotype/CMA result	Age at assessment, mo	Postnatal imaging and intervention	Development
1	Mild	Isolated	NA	72	MRI brain: asymmetrical ventricular system with slightly larger left frontal horn, no focal lesion	Mild global delay
2	Mild	Isolated	Normal	48	MRI brain: ganglioglioma in left temporal lobe	Epilepsy, autism spectrum disorder, speech delay
3	Mild	Isolated	Normal	25	US brain: normal	Mild speech delay
4	Moderate	Isolated (fetal MRI brain: germinal matrix haemorrhage)	Normal	36	MRI brain: previous germinal matrix haemorrhage, small left middle cranial fossa arachnoid cyst	Mild gross motor delay
5	Moderate	Isolated	2p24.1 deletion (VOUS)	48	US brain: mildly dilated lateral ventricles, no other abnormalities	Mild speech and motor delay, autism spectrum disorder
6	Moderate	Isolated	Normal (methylation test after delivery led to diagnosis of Beckwith–Wiedemann syndrome)	29	CT brain: normal	Mild global delay
7	Severe	Isolated	NA	60	US brain: left ventriculomegaly resolved, tiny subependymal cyst at left ventricle	Global delay, epilepsy
8	Severe	Isolated	17p13.3 deletion (pathogenic)	36	MRI brain: mildly prominent ventricles, tiny Rathke's cleft cyst, no other abnormalities	Intellectual disability, speech delay
9	Mild	PLSVC, ARSA	Trisomy 21	25	NA	Mild global delay
10	Mild	Absent cavum septum pellucidum (fetal MRI brain: ischemic changes or cystic encephalomalacia)	Normal (postnatal WES detected homozygous *SCO2* mutation)	14	US brain: periventricular leukomalacia MRI brain: old haemorrhagic insult	Global delay
11	Moderate	TOF	Normal	15 (preterm delivery at 32 weeks)	CT brain: symmetrical periventricular hypodense areas suggest previous ischaemic insult Operation for TOF conducted	Mild global delay
12	Severe	Myelomeningocele (with fetoscopic repair)	Normal	16	MRI brain and spine: persistent low-lying cord and upper syringomyelia, hydrocephalus with aqueduct stenosis VP shunt inserted and operation for cord tethering performed	Gross motor delay

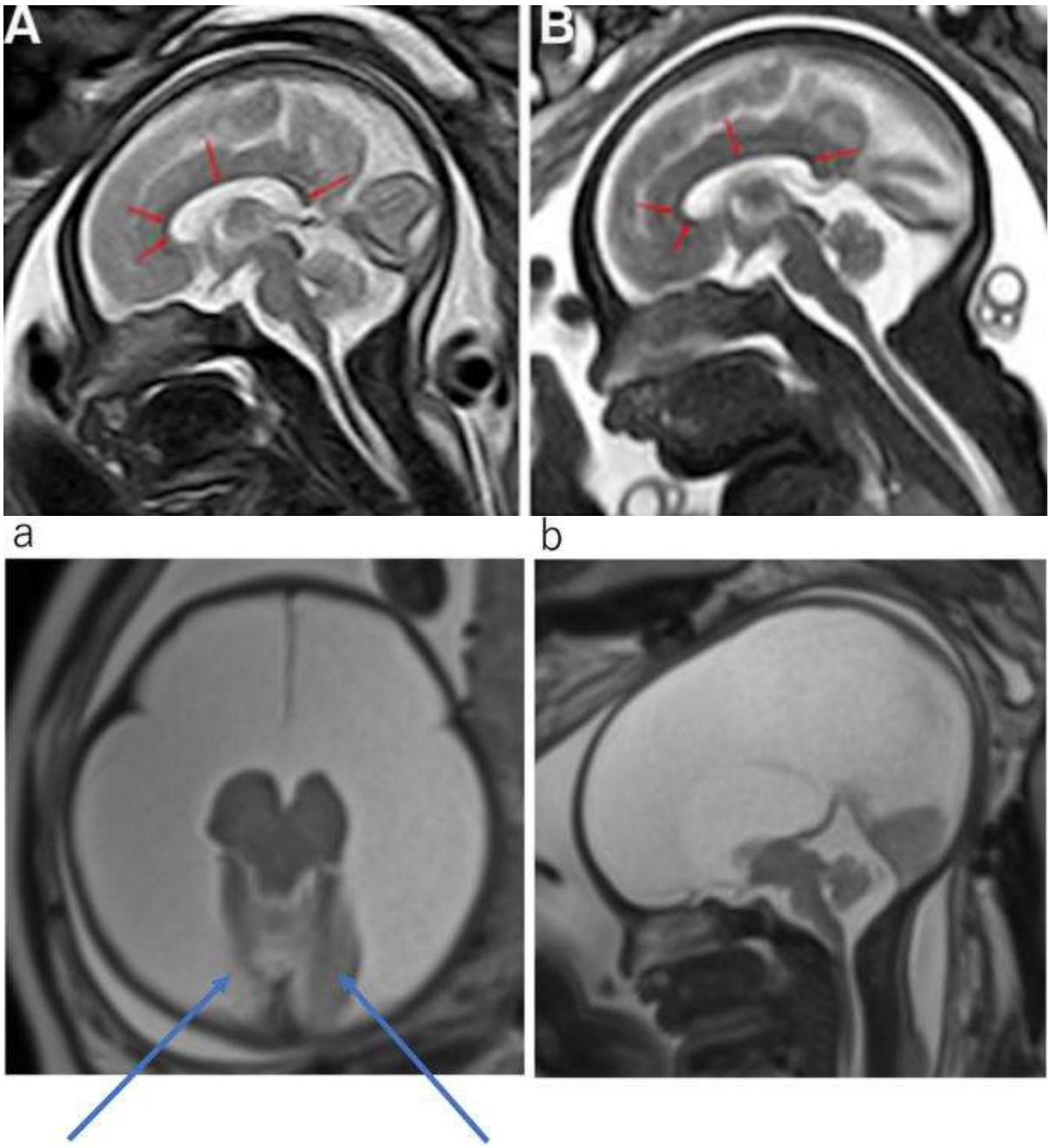
Abbreviations: ARSA = aberrant right subclavian artery; CMA = chromosomal microarray analysis; CT = computed tomography; MRI = magnetic resonance imaging; NA = not available; PLSVC = persistent left superior vena cava; TOF = Tetralogy of Fallot; US = ultrasound; VOUS = variant of uncertain significance; VP = ventriculoperitoneal; WES = whole-exome sequencing

Role of Fetal MRI

Fetal MRI significantly improves detection [2,5] of associated CNS abnormalities not clearly visible on ultrasound.

MRI may identify:
Agenesis of corpus callosum
Cortical malformations
Intracranial hemorrhage
Aqueductal stenosis
Migrational abnormalities
MRI is particularly valuable in:
Progressive VM
Severe VM
Suspected associated anomalies





Genetic Associations

Chromosomal abnormalities are frequently associated [1,5] with fetal ventriculomegaly. Recommended investigations include:

Karyotyping

Chromosomal microarray analysis (CMA)

Whole exome sequencing in selected cases

A recent cohort study demonstrated that CMA significantly increases diagnostic yield compared with conventional karyotyping alone.

Common abnormalities include:

Trisomy 21

Trisomy 18

16p11.2 deletion

22q11.2 abnormalities

Pregnancy Outcomes

Mild Isolated Ventriculomegaly

Most fetuses with isolated mild VM have favorable outcomes [3,4] with normal neurodevelopment. Some cases demonstrate spontaneous resolution during pregnancy.

Moderate and Severe Ventriculomegaly

Moderate and severe VM are associated with:

Neurodevelopmental delay

Seizures

Hydrocephalus

Need for shunting

Increased perinatal mortality

Termination of pregnancy rates are significantly higher in severe VM cases.

Discussion

Recent literature emphasizes [2,6] that ventriculomegaly represents a heterogeneous condition with variable prognosis. Severity, associated anomalies, progression, and genetic findings remain the strongest prognostic indicators. Fetal MRI and CMA have substantially improved prenatal diagnostic accuracy and counseling. However, long-term neurodevelopmental prediction remains challenging because some apparently isolated mild cases may later demonstrate subtle neurological deficits.

Conclusion

Fetal ventriculomegaly has a broad prognostic spectrum. Mild isolated ventriculomegaly generally carries favourable outcomes, whereas severe or non-isolated ventriculomegaly is associated with increased morbidity and mortality. Comprehensive prenatal evaluation [1,2,5] using detailed ultrasonography, fetal MRI, and genetic testing is essential for accurate diagnosis, counselling, and pregnancy management.

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