

The association of fetal choroid plexus cyst with chromosomal abnormalities: A systematic review

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Abstract

Background: Choroid plexus cysts (CPCs) are commonly detected during second-trimester fetal ultrasounds and are often considered benign, particularly in isolated cases. This systematic review aims to look for an association between fetal CPCs and chromosomal anomalies, aiming to provide insights into their diagnostic and prognostic implications.

Methods: Multiple databases were searched to identify studies examining the association of CPCs with chromosomal abnormalities. Included studies were case reports, case series, cross-sectional studies, cohort studies, and case-control studies. Data were extracted independently by two investigators, and statistical analyses were performed to assess the prevalence of chromosomal anomalies in CPC cases.

Results: A total of 12 studies (845 CPC cases) were included in this review. Isolated CPCs (814 cases) were found to have a 2.3% rate of chromosomal abnormalities, whereas CPCs with additional sonographic anomalies (31 cases) showed a significantly higher rate (35.5%) of chromosomal abnormalities ($p < 0.00001$).

Discussion: Isolated CPCs typically do not pose a significant risk for chromosomal abnormalities. However, the presence of associated anomalies needs further evaluation, including genetic counselling and invasive testing. The risk of chromosomal abnormalities in isolated CPC cases is higher than the background risk, thus emphasizing the importance of thorough prenatal screening.

Conclusion: Fetal CPCs with additional anomalies carry a high risk of chromosomal abnormalities, particularly trisomy 18. In isolated CPC cases, a cautious approach involving prenatal counselling and potential invasive testing is recommended, especially in higher-risk pregnancies. Further large-scale prospective studies are needed to refine management protocols and enhance prenatal risk assessment strategies.

Keywords: Choroid plexus cyst; Chromosomal anomaly; Chromosomal abnormalities; Trisomy 18; Trisomy 21; Systematic review

1. Introduction

Choroid plexus cysts (CPCs) are fluid-filled structures within the choroid plexus of the lateral ventricles, often detected during routine second-trimester ultrasound examinations. These cysts are generally considered transient developmental variations, resolving spontaneously by the third trimester in most cases [1]. However, CPCs have garnered clinical significance due to their potential association with chromosomal abnormalities, particularly trisomy 18 and, to a lesser extent, trisomy 21 [2]. The risk of chromosomal anomalies is notably higher when CPCs are

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accompanied by additional sonographic anomalies, raising concerns regarding their diagnostic and prognostic implications [3].

Studies have shown that approximately 30% of fetuses with trisomy 18 exhibit CPCs, compared to about 1% in the general population [3]. Therefore, the clinical management of CPCs remains a subject of debate. While isolated CPCs in low-risk pregnancies are often considered benign with no significant impact on fetal development, their presence in high-risk pregnancies or alongside other anomalies warrants further evaluation. Given these considerations, understanding the likelihood of chromosomal abnormalities in both isolated and non-isolated CPC cases is crucial for refining prenatal counselling and risk assessment strategies.

This systematic review aims to consolidate current evidence regarding the association of fetal CPCs with chromosomal anomalies, highlighting patterns in CPC characteristics, associated anomalies, and pregnancy outcomes. By synthesizing data from multiple study designs, this review assesses the prevalence of chromosomal anomalies in CPC cases, ultimately guiding clinical decision-making in prenatal care.

2. Methods

The study was conducted according to the Preferred Reporting Items for Systematic Reviews and Meta-analysis (PRISMA) guidelines [4].

2.1. Search strategy

Two investigators collected all the literature from sources like PubMed/MEDLINE, PMC (PubMed Central), Google Scholar, Scopus, Cochrane Library, Directory of Open Access Journals (DOAJ), and Clinical Trials Registry (www.ClinicalTrials.gov) using the keywords “fetal” and “choroid plexus cyst”. The search string for the same has been provided in Table 1.

Table 1 Search strings used for different databases

Database	Search string
PubMed	(fetal[Title/Abstract] OR fetus[Title/Abstract] OR foetal[Title/Abstract]) AND ("choroid plexus cyst"[Title/Abstract])
PMC	(fetal[Abstract] OR fetus[Abstract] OR foetal[Abstract]) AND "choroid plexus cyst"[Abstract]
Google scholar	allintitle: "choroid plexus cysts"
Scopus	"choroid plexus cyst"[title], fetal OR fetus OR foetal
Cochrane library	fetal AND "choroid plexus cyst" in All Text
Clinical Trials Register (clinicaltrials.gov)	fetal choroid plexus cyst
OSF registries	Fetal AND "choroid plexus cyst"

All English language articles, including those translated, were gathered with no limit on the date of publication.

2.2. Eligibility criteria

All research articles describing the association of choroid plexus cysts with chromosomal abnormalities have been included for discussion in this review. The types of studies included are case reports, case series, cross-sectional, case-control, and cohort (both prospective and retrospective) studies. All types of review articles, editorials, book chapters, reply letters, and conference abstracts have been excluded for the purpose of this review. Non-human studies as well as non-English articles that have not been translated, have also been excluded. Population, Exposure and Outcome for inclusion have been described below

- Population: All pregnant women undergoing second-trimester fetal anomaly scan
- Exposure: Presence of fetal choroid plexus cyst of any size or number detected in 2nd-trimester anomaly scan

- Outcome: The presence or absence of chromosomal abnormalities like trisomies, monosomies, ploidies, deletions, translocations, etc in fetuses detected by invasive testing or detected in neonates postnatally. There is no time limit for detecting postnatal chromosome abnormalities. Invasive testing can be amniocentesis or chorionic villous sampling.

2.3. Study selection

All search results were imported into Rayyan software [5] for duplicate removal and screening. Investigators obtained training about Rayyan. After duplicate removal, all the papers' titles and abstracts were read by two independent investigators and they opined on study inclusion and exclusion using Rayyan based on the described inclusion and exclusion criteria. During this screening, both were blinded to each other's results. After preliminary screening, further screening of full-text articles was done independently, and the reason for the exclusion of any full-text article was mentioned in Rayyan. After the final study selection, all disputes between both investigators were resolved through discussion.

2.4. Data extraction

Common forms were made in Google Docs spreadsheets, and the following information were collected: author, year, country, study design, number of CPC cases, number of isolated cases and those with other sonographic markers, age of women, size and laterality of choroid plexus cyst, associated sonographic anomalies, invasive testing details, outcome of pregnancy and impression. Data was collected by two investigators independently, and any difference in opinion was resolved with discussion.

2.5. Data synthesis

The results have been discussed qualitatively, analyzing the total number of cases included in this review and the prevalence of chromosomal anomalies. The risk of chromosomal abnormalities in the presence of CPC has been calculated, with a comparative assessment between isolated CPC cases and those with additional sonographic anomalies. The Chi-square test was employed for this comparison, and differences were considered statistically significant if the p-value was less than 0.05. The most frequently observed chromosomal anomaly associated with CPC has been identified, while other factors such as maternal age, CPC size, and laterality have been evaluated in relation to different study designs.

2.6. Quality assessment

Quality assessment of case reports, case series, cross-sectional, case-control and cohort studies were done using Joanna Briggs Institute (JBI) critical appraisal check tools (citation).

3. Results

The PRISMA 2020 flowchart was prepared using the Shiny App [6] (Figure 1).

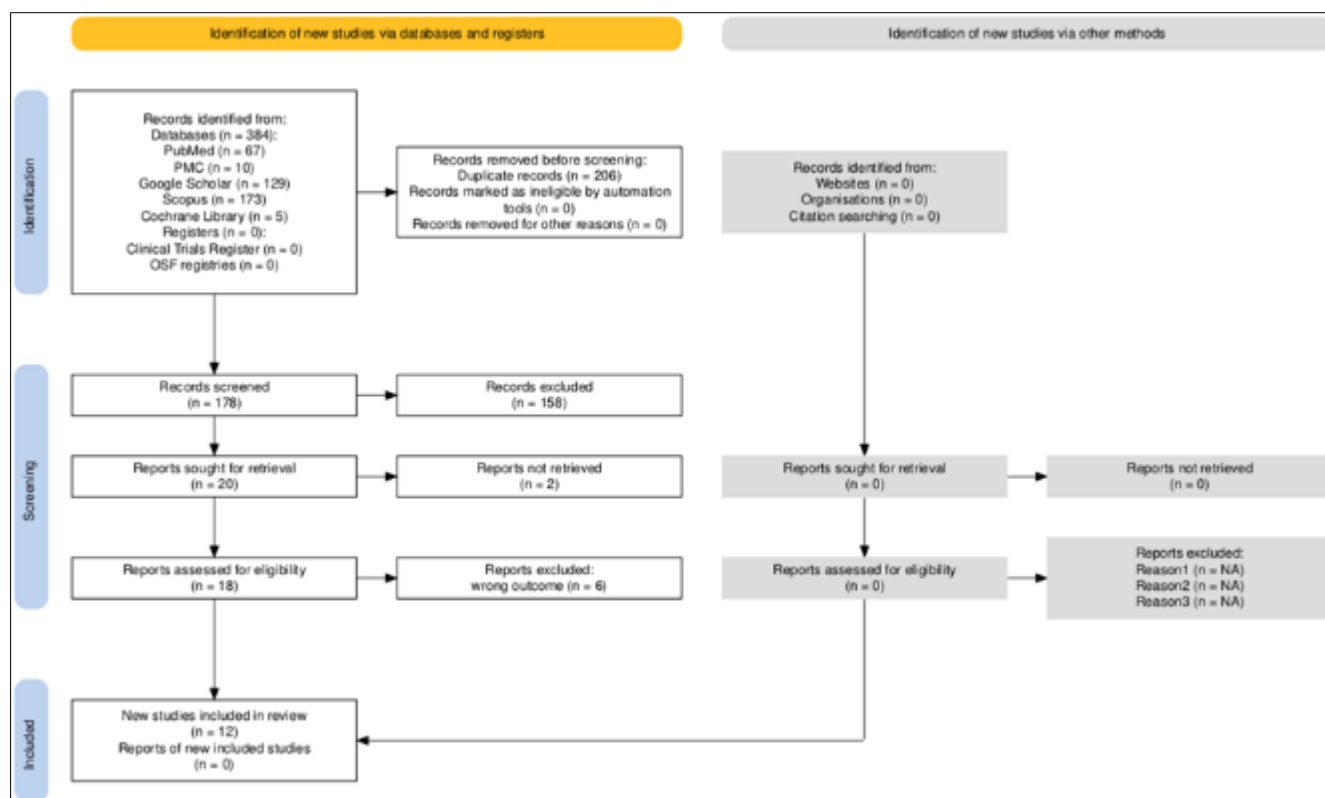


Figure 1 PRISMA flow diagram

A total of 12 studies fulfilling the eligibility criteria after the full-text screening were included in this review. Out of these, there were 5 case reports [7] [8] [9] [10] [11], 2 case series [12] [13], three cross-sectional studies [14] [15] [16], one cohort study [17] and one case-control study [18]. All the study data have been summarized in the table below (Table 2).

Table 2 Summary of included studies

Author, Year, Country	Study Design	Number of Cases	Age of Women	CPC (Single/Multiple)	Size of CPC (mm)	Unilateral/Bilateral	Other Anomalies	Invasive Testing Done	Result of Invasive Test	Outcome of Pregnancy	Comments
Teoh et al., 2004, Singapore [7]	Case Report	1	32 years	Single	NA	Unilateral (Right)	Cerebellar hypoplasia, hypoplastic nasal bone, single umbilical artery	Amniocentesis	46,XX,del(5p14)	Termination due to Cri-du-chat syndrome	-
Sasani et al., 2009, Turkey [8]	Case Report	1	26 years	Single	10mm	Unilateral (Right)	None	Not done	NA	Preterm delivery, normal neonatal karyotype	-
Tayyar & Tayyar, 2016, Turkey [9]	Case Report	1	30 years	Multiple	6mm, 6mm, 4mm	Unilateral	None	Not done	NA	Baby normal at birth	-
Li et al., 2020, China [10]	Case Report	1	29 years	Single	7mm	Unilateral (Right)	IUGR, tricuspid regurgitation, gall bladder not seen	Amniocentesis	46,XX,der(17)t(15;17)(q11.2;q11.2)pat	Termination due to complete trisomy 17p syndrome	-
Chen et al., 2023, Taiwan [11]	Case Report	1	31 years	NA	NA	NA	None	Amniocentesis	47,XX,+21[5]/46,XX[32] (Low-level mosaic trisomy 21)	Delivered at 38 weeks, phenotypically normal at 2-month follow-up	-

Nava et al., 1994, USA [12]	Case Series	211	Mean 30.1 years (15-43)	NA	NA	Unilateral (86), Bilateral (90)	18 cases with anomalies, 4 aneuploidy (3 trisomy 18, 1 trisomy 21)	Amniocentesis in 175, Neonatal karyotyping in 1	Aneuploid in 8 cases	6 terminations due to aneuploidy, 1 fetal death	Amniocentesis should be offered for all CPC cases
Oloyede, 2019, Nigeria [13]	Case Series	8	Median 32 years (27-42)	NA	3-6mm	Unilateral (5), Bilateral (3)	2 cases with anomalies (trisomy 18, trisomy 21)	Amniocentesis in 6, Neonatal karyotyping in 1	Aneuploid in 3 cases (trisomy 21 in 2, trisomy 18 in 1)	NA	-
Irani et al., 2015, Iran [14]	Cross-Sectional	64	Mean 30 years (SD 4)	NA	5-15mm	Unilateral (24), Bilateral (40)	None	Amniocentesis in 8 cases	Normal	Early termination in 4 due to PROM and IUFD	All isolated CPC cases had no chromosomal anomalies
Shah, 2018, India [15]	Cross-Sectional	10	Median 27.5 years	NA	NA	NA	2 cases with anomalies (trisomy 18, clenched hands, growth restriction, polyhydramnios)	Amniocentesis in 1, post-IUD aneuploid detected in 1	Normal	1 IUFD, 1 phenotypically normal birth	Other anomalies should be searched in CPC cases
Masihi et al., 2021, Iran [16]	Cross-Sectional	148	Mean 31 years (SD 4)	NA	NA	NA	None	Not done	NA	Phenotypically normal at birth	Favorable prognosis in isolated CPC cases
Geary et al.,	Cohort Study	84	Median 27 years	NA	NA	NA	6 cases with anomalies (omphalocele,	Amniocentesis in 34	Trisomy 18 in 3 cases	NA	Risk of aneuploidy for isolated

2002, UK [17]			(18-39)				cerebral ventriculomegaly, AV septal defect, absent stomach bubble, diaphragmatic hernia)				CPC is negligible
Wang et al., 2024, China [18]	Case-Control Study	315 CPC cases, 364 controls	Mean 32 years	NA	Mean 7.45mm (3-19mm)	Unilateral (127), Bilateral (188)	None	Amniocentesis in both cases and controls	Abnormal in 13 CPC cases (4.1%) (trisomy 18 in 6, trisomy 21 in 1); 5 controls (1.4%)	NA	Trisomy 18 most common in CPC cases (1.9%); karyotyping & chromosomal microarray should be done

Table 3 Summary of CPC Cases and Chromosomal Anomalies

Category	Number of Cases
Total CPC cases reviewed	845
Isolated CPC cases	814
CPC cases with other associated anomalies	31
Total cases with chromosomal anomalies	30
Trisomy 18 cases	14
Trisomy 21 cases	5
Isolated CPC cases with chromosomal anomalies	19
Trisomy 18	7
Trisomy 21	3
CPC cases with other anomalies and chromosomal anomalies	11
Trisomy 18	7
Trisomy 21	2

A total of 845 cases of choroid plexus cysts (CPCs) were analyzed in this review. Among these, 814 cases were classified as isolated CPCs, while 31 cases had additional associated anomalies. Chromosomal abnormalities were identified in 30 cases (30/845, 3.5%). Specifically, among isolated CPC cases, 19 (2.3%) exhibited chromosomal anomalies, whereas 11 out of 31 cases (35.5%) with other anomalies had chromosomal abnormalities, showing a statistically significant difference (p -value < 0.00001). Trisomy 18 was the most frequently observed chromosomal anomaly, occurring in 14 cases, followed by trisomy 21 in 5 cases.

In the case reports, the maternal age ranged between 26 and 32 years. Four out of five cases had unilateral (right-sided) CPCs, with the largest reported cyst measuring 10mm. Two cases exhibited additional sonographic anomalies, both of which resulted in pregnancy termination due to severe chromosomal abnormalities. In contrast, the remaining three cases without additional anomalies resulted in the birth of phenotypically normal infants.

Both case series were retrospective chart reviews of CPC cases identified during second-trimester ultrasounds, comprising a total of 219 cases. The maternal age ranged from 15 to 43 years. The distribution of unilateral (91 cases) and bilateral CPCs (93 cases) was nearly equal. Additional anomalies were present in 20 cases, of which 4 (20%) were diagnosed with chromosomal abnormalities (aneuploidy). Among the 199 isolated CPC cases, aneuploidy was detected in 5 cases (2.5%).

Three cross-sectional studies discussed a total of 222 CPC cases, with only two cases presenting additional anomalies. Among these, one case underwent invasive testing and was found to have a normal karyotype, while the other case, which showed markers suggestive of trisomy 18, resulted in fetal demise. All isolated CPC cases in these studies were found to be normal at birth.

The cohort study reported 84 CPC cases, with chromosomal abnormalities (trisomy 18) detected in 3 of the 6 cases that had additional anomalies. Meanwhile, the case-control study indicated that chromosomal abnormalities could be identified in approximately 4% of isolated CPC cases using karyotyping and chromosomal microarray analysis. This detection rate was higher compared to cases without any sonographic anomalies.

3.1. Quality assessment

The quality assessment of case reports, case-series, cross-sectional studies, cohort and case-control studies are done using the JBI Critical Appraisal checklist for case reports [19], case-series [20], cross-sectional studies [19], cohort [19] and case-control studies [19], respectively demonstrated in the tables below.

Table 4 Quality assessment of case reports

JBI criteria	Teoh et al., 2004 [7]	Sasani et al., 2009 [8]	Tayyar & Tayyar, 2016 [9]	Li et al., 2020 [10]	Chen et al., 2023 [11]
Were the patient's demographic characteristics clearly described?	Yes	Yes	Yes	Yes	Yes
Was the patient's history clearly described and presented as a timeline?	Yes	Yes	Unclear	Unclear	Yes
Was the current clinical condition of the patient clearly described?	Yes	Yes	Yes	Yes	Yes
Were diagnostic tests or assessment methods and the results clearly described?	Yes	Yes	Yes	Yes	Yes
Was the intervention(s) or treatment procedure(s) clearly described?	Yes	Yes	Not Applicable	Yes	Yes
Was the post-intervention clinical condition clearly described?	Yes	Yes	Yes	Not Applicable	Yes
Were adverse events (harms) or unanticipated events identified and described?	Unclear	Yes	Not Applicable	Not Applicable	Unclear
Does the case report provide takeaway lessons?	Yes	Yes	Unclear	Unclear	Yes
Overall score out of 8	7	8	4	4	7

Table 5 Quality assessment of case-series

JBI Criteria	Nava et al., 1994 [12]	Oloyede, 2019 [13]
Were there clear criteria for inclusion in the case series?	Yes	Yes
Was the condition measured in a standard, reliable way for all participants?	Yes	Yes
Were valid methods used for identification of the condition for all participants?	Yes	Yes
Did the case series have consecutive inclusion of participants?	Yes	Yes
Did the case series have complete inclusion of participants?	Yes	Unclear
Was there clear reporting of the demographics of the participants in the study?	Yes	Yes
Was there clear reporting of clinical information of the participants?	No	Yes
Were the outcomes or follow-up results of cases clearly reported?	Yes	Unclear
Was there clear reporting of the presenting site(s)/clinic(s) demographic information?	Yes	No
Was statistical analysis appropriate?	No	No
Overall score out of 10	8	6

Table 6 Quality assessment of cross-sectional studies

JBI Criteria	Irani et al., 2015 [14]	Shah, 2018 [15]	Masihi et al., 2021 [16]
Were the criteria for inclusion in the sample clearly defined?	Yes	Yes	Yes
Were the study subjects and the setting described in detail?	Yes	Yes	Yes
Was the exposure measured in a valid and reliable way?	Yes	Yes	Yes
Were objective, standard criteria used for measurement of the condition?	Yes	Yes	Yes
Were confounding factors identified and strategies to deal with them stated?	Unclear	No	No
Were strategies to deal with confounding factors stated?	No	No	No
Were the outcomes measured in a valid and reliable way?	Yes	Yes	Yes
Was appropriate statistical analysis used?	Yes	No	Unclear
Overall score out of 8	6	5	5

Table 7 Quality assessment of cohort studies

JBI Criteria	Geary et al., 2002 [17]
Were the two groups similar and recruited from the same population?	Yes
Were the exposures measured similarly to assign people to both exposed and unexposed groups?	Yes
Was the exposure measured in a valid and reliable way?	Yes
Were confounding factors identified?	No
Were strategies to deal with confounding factors stated?	No
Were the groups/participants free of the outcome at the start of the study (or at the moment of exposure)?	Yes
Were the outcomes measured in a valid and reliable way?	Yes
Was the follow up time reported and sufficient to be long enough for outcomes to occur?	Yes
Was follow up complete, and if not, were the reasons to loss to follow up described and explored?	Yes
Were strategies to address incomplete follow up utilized?	No
Was appropriate statistical analysis used?	No
Overall score out of 11	7

Table 8 Quality assessment of case-control studies

JBI Criteria	Wang et al., 2024 [18]
Were the groups comparable other than the presence of disease in cases or the absence of disease in controls?	Yes
Were cases and controls matched appropriately?	Yes
Were the same criteria used for identification of cases and controls?	Yes
Was exposure measured in a standard, valid and reliable way?	Yes
Was exposure measured in the same way for cases and controls?	Yes
Were confounding factors identified?	No
Were strategies to deal with confounding factors stated?	No
Were outcomes assessed in a standard, valid and reliable way for cases and controls?	Yes
Was the exposure period of interest long enough to be meaningful?	Yes
Was appropriate statistical analysis used?	Yes
Overall score out of 10	8

Among all the studies included, only three case reports are of high quality, and the rest of the studies are of moderate quality.

4. Discussion

The findings of this review provide critical insights into the association between choroid plexus cysts (CPCs) and chromosomal abnormalities, particularly trisomy 18 and trisomy 21. Among the 845 cases analyzed, the vast majority (814 cases) were isolated CPCs, with a smaller subset (31 cases) presenting additional anomalies. The significant difference in the prevalence of chromosomal abnormalities between isolated CPC cases (2.3%) and those with associated anomalies (35.5%) highlights the importance of evaluating CPCs in the context of other sonographic markers. The statistical significance (p -value < 0.00001) further strengthens this finding, reinforcing the need for careful prenatal assessment when CPCs are detected.

4.1. Clinical Implications of CPCs

The low prevalence of chromosomal abnormalities in isolated CPC cases aligns with previous studies suggesting that isolated CPCs are generally benign findings. However, the presence of additional anomalies substantially increases the likelihood of aneuploidy, particularly trisomy 18. Therefore, a thorough ultrasound evaluation is necessary to rule out other structural abnormalities that could indicate a higher risk of chromosomal defects. The case-control study included in this review further supports this notion by demonstrating that chromosomal microarray analysis can detect abnormalities in approximately 4% of isolated CPC cases, which is a slightly higher prevalence than previously reported.

4.2. Impact of Study Design on Findings

The review includes various study designs, such as case reports, case series, cross-sectional studies, a cohort study, and a case-control study, offering a comprehensive perspective on CPCs. Case reports primarily provided detailed clinical insights into individual cases, demonstrating variability in CPC presentation and outcomes. The case series and cross-sectional studies helped establish broader epidemiological patterns, confirming that isolated CPCs are largely associated with favorable pregnancy outcomes. The cohort and case-control studies provided valuable comparative analyses, reinforcing the role of additional sonographic markers in risk assessment.

4.3. Prenatal Counselling and Management

While isolated CPCs alone should not be a cause for undue concern, the detection of additional anomalies warrants a more detailed evaluation, including genetic counselling and potential invasive testing such as amniocentesis. The decision to proceed with further testing should be individualized, taking into account maternal age, family history, and

other ultrasound findings. The fact that most isolated CPC cases resulted in the birth of phenotypically normal infants provides reassurance to parents and clinicians alike.

4.4. Quality assessment of included studies

Though the case reports were of moderate to high quality, both case series and cross-sectional studies lacked proper inclusion of study participants. None of the analytical studies had an appropriate inferential statistical approach, and most of them only provided descriptive data. Confounders have not been addressed, and therefore, studies suffer from methodological weakness.

4.5. Limitations and Future Directions

Despite the comprehensive nature of this review, some limitations must be acknowledged. First, the sample sizes in individual studies varied, which may have influenced the overall prevalence rates of chromosomal abnormalities. Additionally, retrospective study designs, particularly in case series, may introduce selection bias, limiting the generalizability of findings. Also, the quality of the included studies was mostly moderate, and none of the analytical studies were of high quality, which might provide biased results. Future research should aim to incorporate larger, multicenter cohort studies and prospective studies with long-term follow-up to further elucidate the clinical significance of CPCs.

5. Conclusion

Fetal CPCs with other structural anomalies have a very high risk of chromosomal abnormalities, particularly trisomy 18, for which genetic testing must be offered. In those with isolated CPCs, the decision-making is tricky as though its prognosis is favourable in most, yet there is a chance of chromosomal anomaly with a risk higher than the background risk. Therefore, invasive testing may be considered based on history and patient willingness. The use of both karyotyping and chromosomal microarray analysis may be offered as it has been shown to have a higher detection rate. Ultimately, a thorough sonographic evaluation followed by proper pre-natal counselling with genetic testing may be the best management protocol for tackling a case of choroid plexus cyst.

Compliance with ethical standards

Disclosure of conflict of interest

The authors declare no conflict of interest for this article.

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