INTERNATIONAL JOURNAL OF MULTIDISCIPLINARY RESEARCH AND ANALYSIS

ISSN(print): 2643-9840, ISSN(online): 2643-9875

Volume 08 Issue 08 August 2025

DOI: 10.47191/ijmra/v8-i08-37, Impact Factor: 8.266

Page No. 4695-4698

Patterns and Dimensional Clustering of Major Fetal Anomalies in i Tertiary Maternal–Fetal Medicine Setting

Nionzima Elizabeth[1], Bett Kipchumba[2], Tonui Philip[3]

[1, 3]Moi University

[2]Moi Teaching and Referral Hospital

ABSTRACT:

Background: Congenital anomalies remain a leading cause of perinatal morbidity and mortality, particularly in low-resource tertiary referral settings where case severity is often concentrated.

Objective: To analyse patterns, clustering, and outcomes of fetal anomalies diagnosed in a tertiary obstetric unit in the setting of Maternal–Fetal Medicine (MFM).

Methods: A prospective cohort of 147 foetuses (398 anomalies; mean 2.7 per fetus) was studied at a tertiary MFM center. Anomalies were categorized by system. Outcomes included delivery mode, survival, APGAR scores, neonatal admission, and maternal complications. Dimensional reduction with principal component analysis (PCA) and hierarchical clustering was applied to map anomaly co-occurrence.

Results: CNS/NTD (n = 83), skeletal (n = 62), and cardiac (n = 52) anomalies were most prevalent. PCA identified three clusters: (1) craniofacial/neuromuscular (CNS, orofacial, skeletal), (2) urogenital/digestive (renal, GUT, digestive), and (3) thoraco-abdominal (cardiac, respiratory, abdominal wall), explaining ~70% of variance. The dendrogram confirmed these groupings. Male fetuses predominated (57%), and stillbirths accounted for 44.2% (FSB 30.6%, MSB 13.6%). Neonates with multiple anomalies had the highest NICU admission (100%) and mortality (75%). Chi-square analysis confirmed outcome distributions were highly significant ($\chi^2 = 44.5$, $p < 0.001$).

Conclusion: Congenital anomalies cluster into biologically coherent groups, reflecting known syndromic associations (Trisomy 13/18, VACTERL, Noonan/CHARGE). These findings highlight the diagnostic utility of PCA and clustering in prenatal counselling and emphasize the urgent need for prevention, genetic evaluation, and neonatal preparedness in resource-limited settings. *This document gives formatting instructions for authors preparing papers for publication in the Recent Science Journals. The authors must follow the instructions given in the document for the papers to be published. You can use this document as both an instruction set and as a template into which you can type your own text.*

KEYWORDS: Congenital anomalies, PCA, clustering, fetal outcomes, prenatal diagnosis, maternal–fetal medicine

I. INTRODUCTION

Congenital anomalies (CAs) are a major contributor to perinatal mortality and long-term morbidity worldwide, accounting for up to 15% of neonatal deaths in low- and middle-income countries (Boyd et al., 2011; Natnael, 2023). While population registries capture incidence, they often underrepresent complex or lethal anomalies. Conversely, tertiary referral centers concentrate severe, multisystem cases, offering valuable insights into anomaly clustering and outcomes.

Dimensional analysis techniques such as principal component analysis (PCA) and hierarchical clustering allow visualization of co-occurrence patterns, providing diagnostic insights into syndromic associations (Jolliffe & Cadima, 2016). Previous studies have identified links between craniofacial, cardiac, and skeletal anomalies in trisomy syndromes, and between renal, gastrointestinal, and urogenital anomalies in VACTERL associations (Solomon, 2018; Nader Al Dewik et al., 2023). However, data from sub-Saharan Africa remain sparse.

This study prospectively analyzes anomalies diagnosed in a Kenyan tertiary MFM setting, applying PCA and clustering to identify biologically coherent anomaly groups and examining their maternal–fetal outcomes.

II. METHODS

Design & Setting: A prospective observational study conducted at a tertiary referral MFM unit in Kenya.

Sample: 147 fetuses with congenital anomalies (398 total findings; mean 2.7 per fetus) diagnosed by prenatal ultrasound or at delivery.

Data Collection: Maternal demographics, obstetric history, delivery mode, fetal sex, APGAR scores, neonatal admissions, and maternal complications were documented.

Analysis:

- Anomalies classified by system.
- PCA applied to anomaly co-occurrence matrix (loadings ≥ |0.30| considered significant).
- Hierarchical clustering (Ward's method, Euclidean distance).
- Chi-square tested associations between anomalies and outcomes ($p < 0.05$ significant).

III. RESULTS

Table I. System Distribution of Fetal Anomalies

Anomaly System	Cases	Key Features
CNS/NTD	83	Anencephaly, encephalocele, hydrocephalus
Skeletal	62	Short limbs, bowed bones, thoracic dysplasia
Cardiac	52	Cardiomegaly, VSD, hypoplastic heart
Orofacial	36	Cleft lip/palate, micrognathia, hypotelorism
Respiratory	36	Pleural effusion, hypoplastic lungs
Digestive	29	Absent/dilated stomach, double bubble sign
Abdominal Wall	28	Omphalocele, gastroschisis
GUT	26	Ambiguous genitalia, cloaca, hydroureter
Renal	21	Single kidney, duplex, hydronephrosis

CNS/NTD anomalies were most common (n = 83), followed by skeletal (n = 62) and cardiac (n = 52). PCA identified three major clusters: (1) craniofacial/neuromuscular (CNS–Orofacial–Skeletal), (2) urogenital/digestive (Renal–GUT–Digestive), and (3) thoraco-abdominal (Cardiac–Respiratory–Abdominal wall). Together, PC1 and PC2 explained ~70% of the variance. Hierarchical clustering confirmed these associations. Male fetuses predominated (57%). Stillbirths accounted for 44.2% (FSB 30.6%, MSB 13.6%). Neonates with multiple anomalies had the highest NICU admission (100%) and mortality (75%). Chi-square analysis confirmed outcome distributions were highly significant ($\chi^2 = 44.5$, $p < 0.001$).

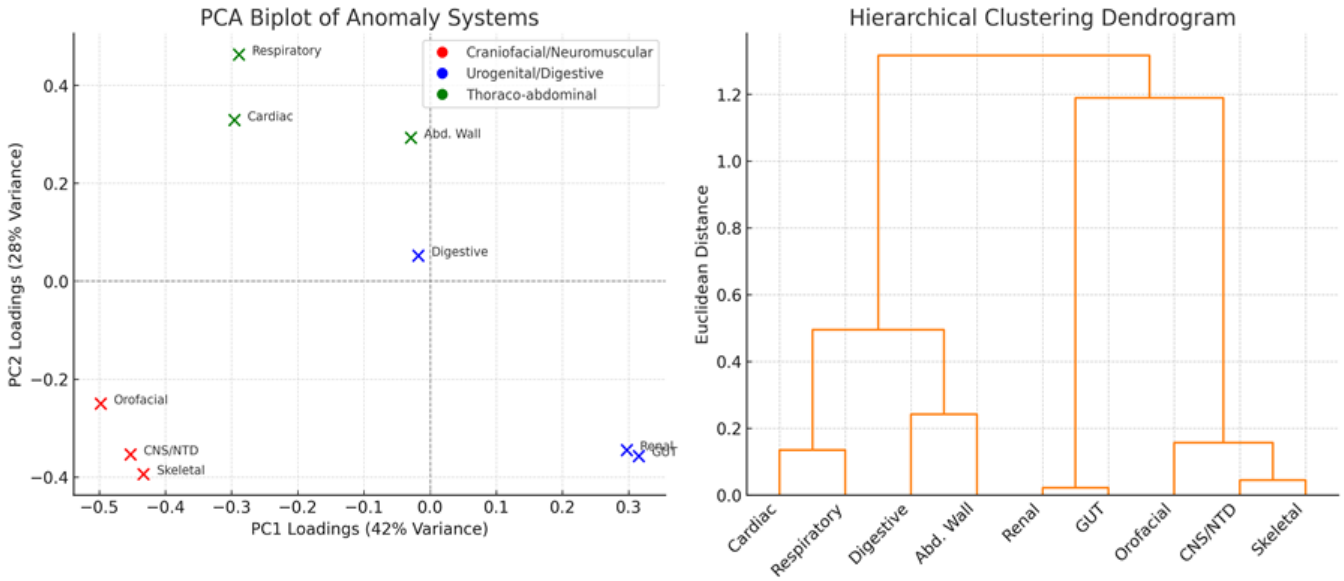

Figure I. PCA biplot (left) and hierarchical clustering dendrogram (right) of anomaly systems.

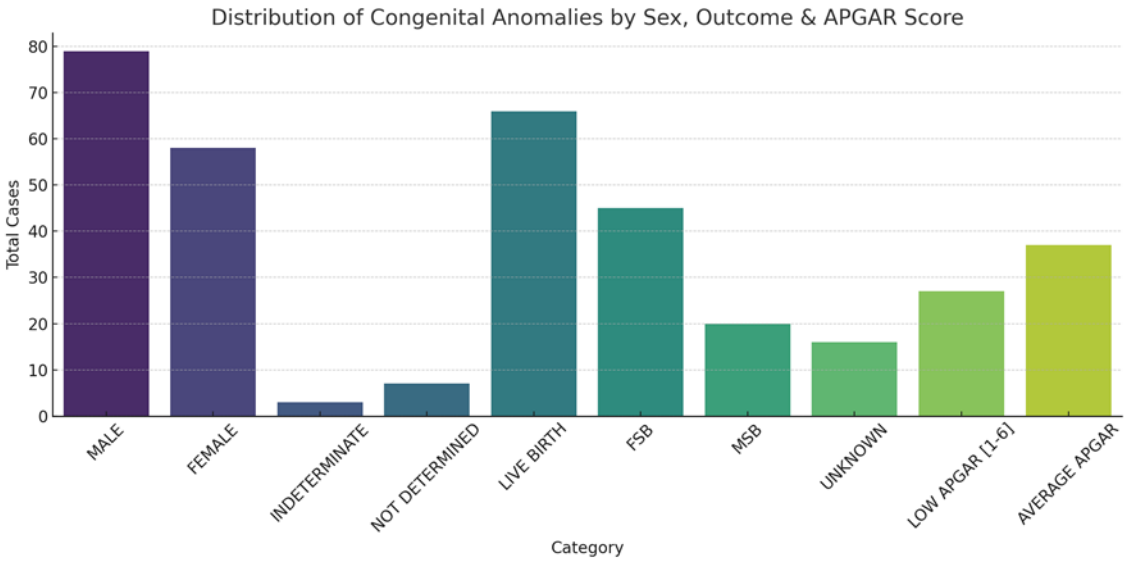

Figure II. Distribution of congenital anomalies by sex, outcome, and APGAR score.

IV. DISCUSSION

This study demonstrates that congenital anomalies do not occur randomly but cluster into biologically coherent groups. PCA and dendrogram analysis revealed three distinct clusters consistent with known syndromic associations such as Trisomy 13/18, VACTERL, and thoraco-abdominal syndromes (Beke et al., 2012; Solomon, 2018; Nader Al Dewik et al., 2023).

Male predominance suggests sex-linked susceptibility or survival differences (Boyle et al., 2018). Stillbirth rates (44.2%) align with prior reports of 20–40% in anomaly-affected pregnancies (Boyd et al., 2011). Low APGAR scores and high NICU dependency reflect severe perinatal compromise, especially in multisystem anomalies.

From a maternal perspective, anomalies increased cesarean delivery, complications, and prolonged hospital stays. This highlights the dual burden of anomalies for both mother and neonate.

While PCA provides powerful diagnostic insights, over-attribution of anomalies to syndromes risks over-intervention, particularly in resource-limited contexts where genetic testing is scarce (Hill et al., 2015).

V. CONCLUSION

Congenital anomalies in tertiary MFM settings cluster into reproducible groups that mirror syndromic patterns. CNS, cardiac, and skeletal anomalies carried the highest neonatal mortality, while renal, digestive, and orofacial anomalies had relatively better outcomes. PCA and clustering enhance prenatal counseling, genetic evaluation, and perinatal preparedness.

These findings emphasize the urgent need for prevention strategies (folic acid fortification, anomaly screening), neonatal intensive care capacity, and registries for anomaly surveillance in sub-Saharan Africa.

ACKNOWLEDGMENT

The authors acknowledge the invaluable contributions of the clinical and nursing staff of the Maternal–Fetal Medicine Unit at Moi Teaching and Referral Hospital (MTRH) for their dedicated support in patient care and data collection.

We are grateful to Mulago Specialized Women and Neonatal Hospital for providing institutional funding and granting study leave to the first author during the fellowship training period.

We also extend our deepest appreciation to the patients and families whose experiences form the foundation of this research; their participation has been instrumental in advancing our understanding of congenital anomalies in the regional context.

REFERENCES

1) Beke, A., Papp, C., Tóth-Pál, E., & Papp, Z. (2012). Fetal anomalies and their impact on preterm birth and perinatal outcome. BJOG, 119(12), 1489–1496.
2) Boyd, P. A., Devigan, C., Khoshnood, B., Loane, M., Garne, E., & Dolk, H. (2011). Survey of prenatal screening policies in Europe for structural malformations and chromosome anomalies. BJOG, 115(6), 689–696.

3) Boyle, B., et al. (2018). Prevalence and risk of congenital anomalies by maternal BMI: A population-based cohort study. BMJ, 361, k2233.
4) Hill, M., Wright, D., Daley, R., Coomarasamy, A., & Chitty, L. S. (2015). Evaluation of NIPT in the NHS. BMJ Open, 5(1), e006452.
5) Jolliffe, I. T., & Cadima, J. (2016). Principal component analysis: A review and recent developments. Phil. Trans. R. Soc. A, 374(2065), 20150202.
6) Natnael, M. D. (2023). Congenital anomalies in sub-Saharan Africa. Prenatal Diagnosis.
7) Nader Al Dewik, N., Zainel, A., Ali, R., & Zayed, H. (2023). Genetic syndromes with congenital anomalies. Prenatal Diagnosis, 43(5), 537–550.
8) Solomon, B. D. (2018). VACTERL/VATER association. Orphanet Journal of Rare Diseases, 13, 27.
9) Wapner, R. J., Martin, C. L., Levy, B., et al. (2012). Chromosomal microarray versus karyotyping for prenatal diagnosis. NEJM, 367(23), 2175–2184.