Clinical, Imaging, and Genetic Spectrum of Polymicrogyria in Indian Children

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INTRODUCTION

- Polymicrogyria (PMG) is one of the commonest cortical malformations associated with varied clinical presentations.
- It also has heterogenous radiological features with various subtypes with genetic correlation,[1] yet large studies defining the clinical, imaging and genetic heterogenicity are lacking among Indian children.
- Herein, we describe the clinical, imaging and genetic spectrum of PMG in pediatric population, and classify polymicrogyria in terms of pattern and subtypes.

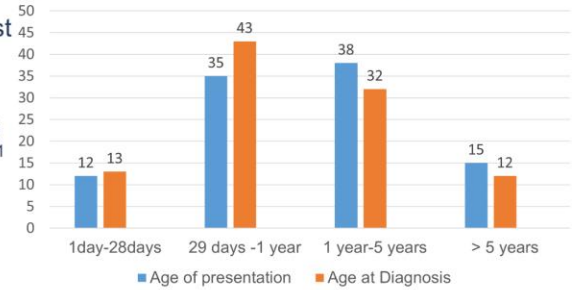
OBJECTIVES

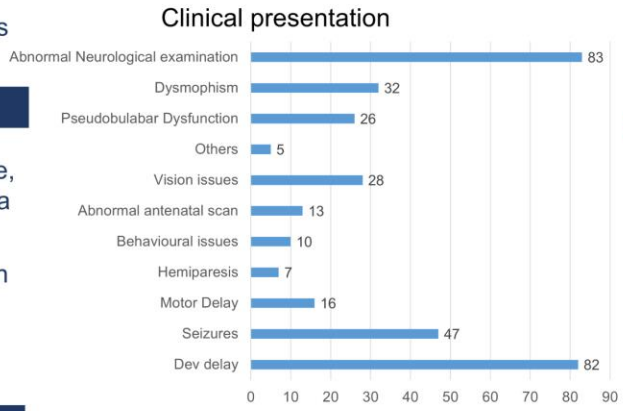
- **Primary Objective :** To describe clinical profile, imaging findings of children with polymicrogyria
- **Secondary Objective:**
- a) To classify polymicrogyria in terms of pattern and subtype
- b) To describe genetic spectrum of polymicrogyria

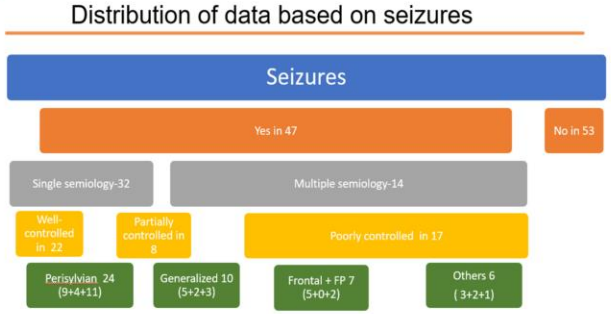
MATERIAL AND METHODS

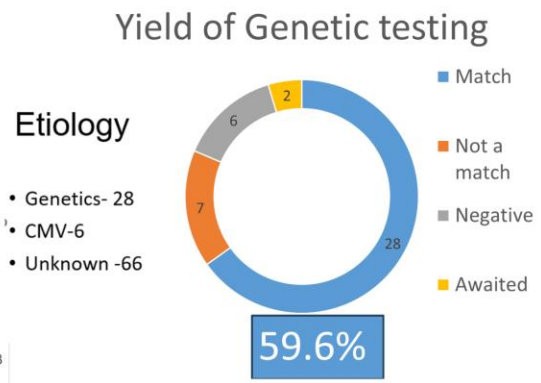
- **Type of study:** Descriptive, cross-sectional, observational study
- **Sample size**:100
- **Study Period:** Oct 2021 to Oct 2023
- **Study place:** Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad.
- **Inclusion Criteria:** Children aged 1 day to 16 years with a radiological diagnosis of polymicrogyria.

RESULTS



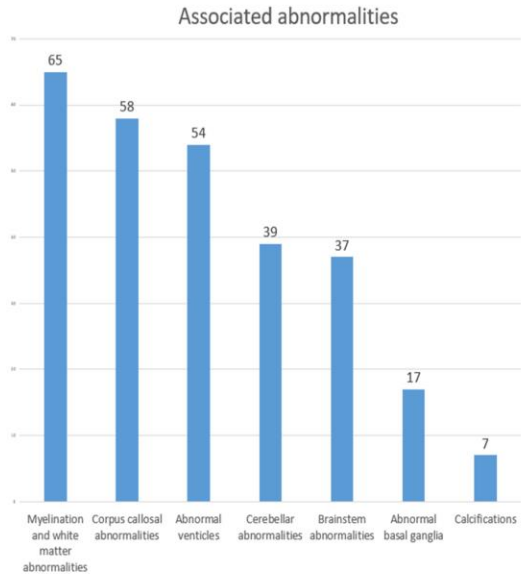


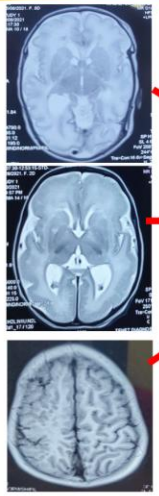




Genes Identified

DYNC1H1
NSD1
PONGNT
COL4A2
RAB3GAP2
MAP3K1
ACOX
KIFBP
HSD17B4
GABBR2
ATP1A3
TRIM2
SMPD4
SZT2
ADGRG1
CCDN2
OSGEP
B3GALNT2
SNAP29
PACS2
CASK
GRIN2B
4p.16.1





Location	Subtypes	Number
Perisylvian (45)	Perisylvian only	13
	Perisylvian with others	32
Generalized (28)	Normal WM	12
	Abnormal WM	16
Frontal (15)	Frontal only	5
	Frontoparietal	10
Others (12)	Temporal/occipital/parietal/vermis	12

ADGRG1 mutation was found in 7 patients and all had *generalized polymicrogyria* and brainstem changes

CONCLUSION

- Most common presentation is developmental delay followed by seizures with most common age of presentation is between 1year to 5 years.
- Perisylvian PMG is the commonest radiological pattern followed by generalized polymicrogyria.
- ADGRG1 is the commonest genetic cause of PMG in Indian children

REFERENCES

1.Stutterd CA, Brock S, Stouffs K, et al. Genetic heterogeneity of polymicrogyria: study of 123 patients using deep sequencing. Brain Commun. 2020;3(1):fcaa221. Published 2020 Dec 26. doi:10.1093/braincomms/fcaa221