Aicardi-Goutieres Syndrome: A single center cohort of thirty-eight genetically confirmed cases from Southern India

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INTRODUCTION

•Aicardi-Goutieres syndrome (AGS) is a rare neurogenetic disorder characterised by inflammation

•Mode of inheritance : autosomal recessive inheritance, and in a few instances due to autosomal dominant variety or de novo mutation [1].

•It is frequently misdiagnosed and treated as congenital infection (toxoplasma , CMV, rubella, etc.) , pyrexia of unknown origin, hypoxic ischemic encephalopathy, etc. [2]

OBJECTIVES

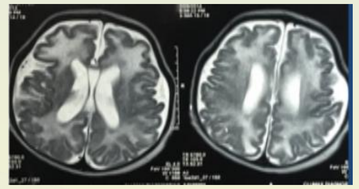
To determine the clinical, radiological and laboratory profile including molecular spectrum of AGS.

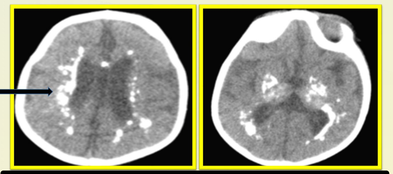
MATERIALS AND METHODS

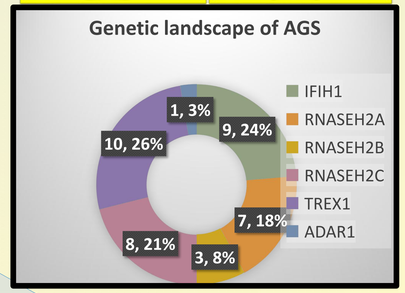
This is a retrospective chart review of children with genetically confirmed AGS at a tertiary care institute in Southern India over last six years

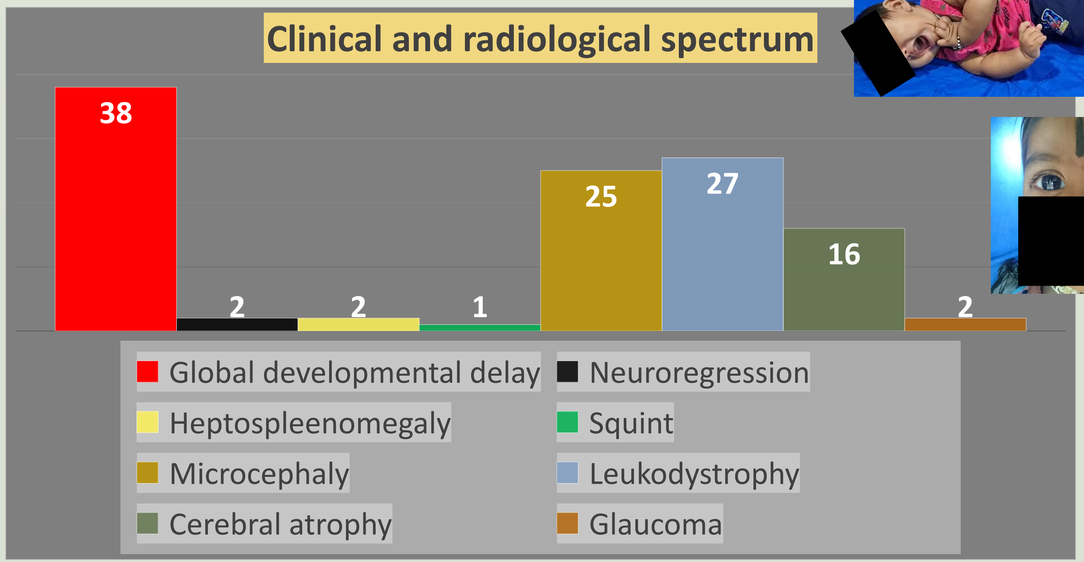
RESULTS

- Total 38 children (25 males and 13 females)
- Mean age of diagnosis being at around 3.4 years
- Mean age of presentation being at around 1 years, indicating time lag of 2.4 years .
- All 38 of them had global developmental delay with spastic quadriparesis with epilepsy
- 4 of the patients did not have any intracranial calcification









CONCLUSION

GDD with without regression with seizures with intracranial calcifications are the most common manifestations.

Atypical features include glaucoma and craniosynostosis.

CSF findings were normal in all cases

CT brain or SWI/GWI of MRI of brain should be considered in all suspected cases of AGS to look for calcification for early diagnosis.

Most common mutation wasTREX1 gene.

Significant time lag between age at presentation and age of diagnosis is seen indicating lack of characteristic clinical features and affordable and easy to use diagnostic methods.

Whole exome sequencing helps in early diagnosis and avoid unnecessary investigation.