# When Heart Tricks the Brain: Recurrent episodes of stroke revealing Concealed Congenital Long QT1 Syndrome

**Presenting Author- DR. S. HEMA KUMAR[1], DR. P. R. SOWMINI[2], DR. S. SAKTHI VELAYUTHAM[2], DR. K. MALCOLM JEYARAJ [2], DR. KRISHNA KUMAR[2], DR. V. KANNAN[3], DR. S. VELUSAMY[3], DEPARTMENT OF NEUROLOGY, STANLEY MEDICAL COLLEGE AND HOSPITAL, CHENNAI.**

1-Resident, 2-Assistant Professor, 3-Professor

## AIMS

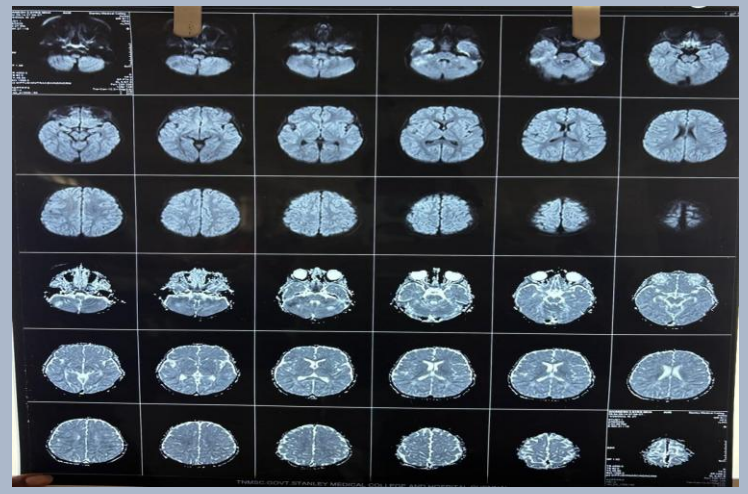
To present a rare case of Concealed Congenital Long QT1 Syndrome manifesting as recurrent syncopal attacks on exertion which initially misdiagnosed as seizure and initiated on ASM. This case highlights the importance of need for comprehensive diagnostic work up and should always look for a treatable etiology.

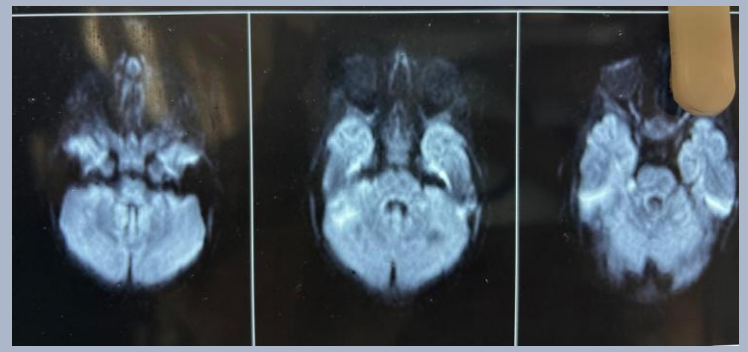
## *CASE HISTORY:*

***Here we present a case of 2½ year old child with an unremarkable birth and developmental history. History of camphor induced seizure(GTCS) at 1 year of age, presented with recurrent paroxysmal events that were diagnosed as seizures and treated with antiseizure medication. Despite therapy, the episodes persisted. Detailed history and clinical examination later revealed that these were syncopal attacks rather than seizures. The child experienced brief episodes of giddiness lasting 2-3 minutes, accompanied by swaying while attempting to rise from sitting or supine position, lethargic and occasional vomiting. These episodes were triggered by exertional activity like jumping of the bed, dancing and playing. Between episodes, the child remained completely normal with no focal neurological deficits and normal CNS examination. During the event, examination revealed bilateral gaze evoked nystagmus and transient postural instability, but later the child recovered spontaneously in minutes.***

## METHODS:

- Clinically child is diagnosed with seizure initially with history of camphor induced seizure and empirically started on anti-seizure medication.
- Despite on medication, child developed similar episodes of giddiness and MRI brain showed acute infarct in right superior cerebellar hemisphere, left caudate and multiple chronic lacunar infarcts in bilateral cerebellar hemispheres. EEG was done which was normal.
- Pediatric stroke work up: Arteriopathies and cardiac disease are being the most common causes, CT-Angiogram and dynamic MR Angiogram were normal. ECG and 2D ECHO were normal. Further thrombophilia, vasculitis and connective tissue disease work up was normal.
- Metabolic disorders like mitochondrial were suspected and serum lactate was done which is mildly elevated (38mg/dl).
- Genetic analysis: Whole exome sequence with mitochondrial DNA analysis was done, which showed heterozygous pathogenic variant, KCNQ1 suggestive of Congenital Long QT1 Syndrome.





## RESULTS:

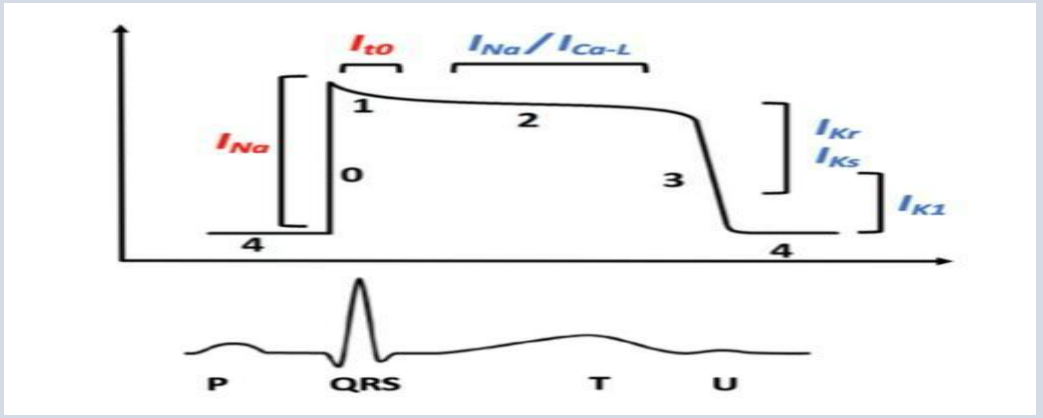
- **Pathogenic mutation for Long QT1 Syndrome with stress induced syncope accounts for Modified Schwartz Score of 5.5. ( >3.5 suggest LQTS as per Elia Balestra et al 2024)[2].**
- **Routine ECG and ECHO can be normal in concealed Long QT1 Syndrome[1].**
- **Ambulatory Holter monitoring or Exercise stress test may reveal prolonged corrected QT interval(≥ 480ms during 4rth minute of recovery from exercise stress test)[2].**

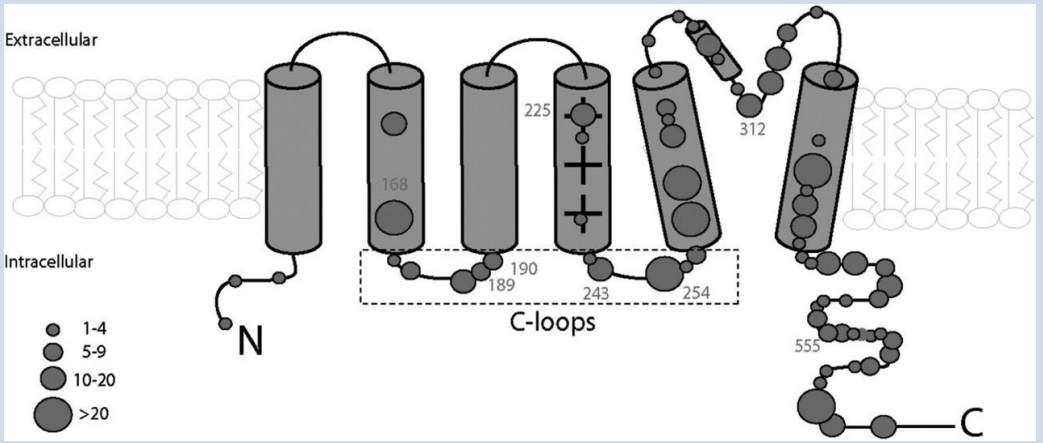
**Summary of Variants**

| Gene and Transcript | Exon/Intron Number | Variant Nomenclature [Variant depth/ Total depth] | Zygosity | Classification | OMIM Phenotype | Inheritance |
|---|---|---|---|---|---|---|
| KCNQ1 (NM_000218.3) | Exon 4 | c.613G>A p.Val205Met [36x/88x] | Heterozygous | Pathogenic | Long QT syndrome 1 | Autosomal dominant |

## DISCUSSION:

- Long QT1 syndrome is an inherited **primary arrythmia syndrome** that may present with **malignant arrythmia** and **risk of sudden cardiac death**[1]
- Cardinal symptoms being palpitations, syncope and anoxic seizures secondary to ventricular arrythmias, classically '**torsades de pointes'**
- Concealed or silent LQTS1 due to **loss of function mutation in delayed rectifier channel of potassium(Iks)**, surface ECG can be normal and patient develop symptoms only during exertion[1]
- This case emphasizes that patients presenting with recurrent syncopal attacks on exertion with **normal ECG doesn't rule out cardiac etiology**
- Hence Ambulatory Holter monitoring or Exercise stress test is warranted in cases of unexplained exertional syncopal attacks in both extremes of age group
- Clinical suspicion and early diagnosis will prevent morbidity and mortality
- Child is treated with propranolol and he is asymptomatic.




## CONCLUSION:

**Syncope and Seizures are great mimickers of each other. Therefore, a thorough history and detailed clinical examination along with supportive investigations are essential to differentiate. A normal Echocardiogram and ECG do not necessarily rule out cardiac etiology. Early diagnosis of LQTS and appropriate management are crucial for reducing morbidity and mortality, ultimately improving patient outcome. Always look for a treatable etiology.**


REFERENCES

1. Wallace E, Howard L, Liu M, O'Brien T, Ward D, Shen S, Prendiville T. Long QT Syndrome: Genetics and Future Perspective. Pediatr Cardiol. 2019 Oct;40(7):1419-1430.
2. Balestra E, Bobbo M, Cittar M, Chicco D, D'Agata Mottolese B, Barbi E, Caiffa T. Congenital Long QT Syndrome in Children and Adolescents: A General Overview. Children (Basel). 2024 May 11;11(5):582.
3. Gu C, Zhang Y, Sun M, Zhou F, Cai C. Clinical and genetic characteristics of 18 cases with suspected congenital long QT syndrome: a retrospective cross-sectional study. Heart Mind. 2024 Apr-Jun;8(2):117-23.