

Asystolic pause as a mechanism of sudden death in Rett syndrome: Case presentation and literature review

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Objectives: To describe a case of asystole in a patient with Rett Syndrome and review medical literature on Rett syndrome and associated cardiac comorbidities, including proposed mechanisms of unexplained sudden death.

Background: Exclusively affecting girls, Rett syndrome is associated with mutations in the MEPC2 gene on the X chromosome and presents as progressive loss of cognitive and motor development after initially normal development. Previous studies have reported annual mortality rate of 1.2% in patients with Rett Syndrome, of which 26% were sudden and unexplained. Previously proposed mechanisms of sudden death in Rett syndrome patients include seizures, autonomic dysfunction, respiratory abnormalities, and arrhythmias.

Case Report: A 16-year-old girl with Rett syndrome with new-onset sleep disturbance underwent cardiac arrhythmia monitoring with ZioPatch to exclude cardiac arrhythmias. A 31.2 second asystolic pause followed by three escape beats and two subsequent asystolic pauses of 6.1 and 3.7 seconds were documented. Sinus rhythm with normal repolarization was noted before and after this event. The asystolic event was not associated with sleep disorder, but occurred on a day when she had a seizure. The remaining event recording showed no significant tachycardia, bradycardia, or asystolic events.

Previous ECGs and Holter showed sinus with normal to borderline-prolonged QTc (longest QTc = 0.467s). Given the prolonged asystolic event, she underwent single chamber pacemaker implantation. She has required <0.1% pacing with no additional arrhythmias documented.

Conclusions: Despite increased risk of sudden death events, autonomic disarray, and prolonged repolarization, there is no recommendation for prolonged cardiac arrhythmia monitoring in Rett syndrome patients. Based on this case and aforementioned comorbidities, it seems reasonable to consider longer-term cardiac monitoring, such as non-invasive event monitor or minimally-invasive implantable loop recorder to better characterize cardiac rhythm disturbances and identify those individuals who would benefit from a therapeutic device implantation to avoid life-threatening bradycardia and asystole.

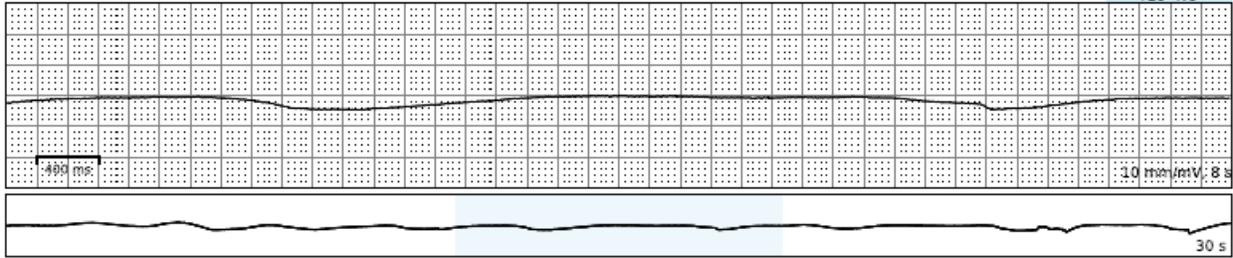
1

Longest Pause

11/07/15 01:40:51pm

Duration:
31.2 s (2 bpm)

Pt Triggered?
 YES NO



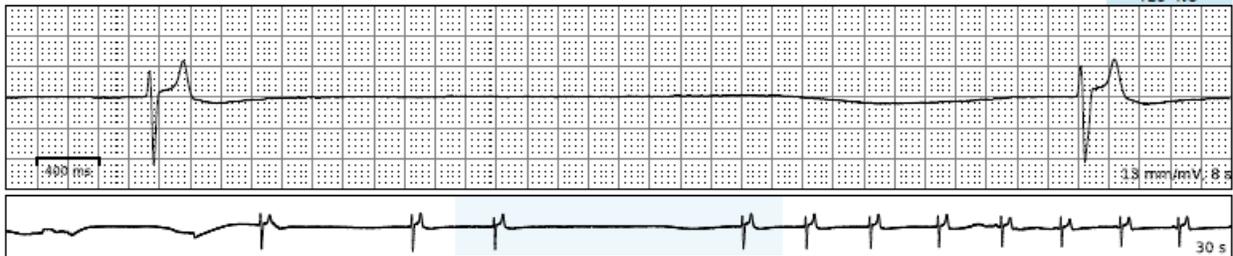
2

Second Longest Pause

11/07/15 01:41:28pm

Duration:
6.1 s (10 bpm)

Pt Triggered?
 YES NO



3

Third Longest Pause

11/07/15 01:41:22pm

Duration:
3.7 s (16 bpm)

Pt Triggered?
 YES NO

