Case Report

Diseases that masquerade neurology

Malcolm Gladwell introduces the concept of 'snap judgement' or as he says 'the power of thinking without thinking' in his book: Blink. Our adaptive unconscious is that part of our brain, which leaps to a conclusion by way of thin-slicing much before our logical mind has completed its analysis and processing. On some occasions, I have experienced this; it has so happened that before, during or immediately after completing a consultation I have written down the diagnosis and not even thought about it! I am not indulging into self-praise here. I want to narrate to my reader one such incidence of 'spot diagnosis' as we call it and how our unconscious and conscious mind work towards managing our patients.

Presentation

An 8 year old boy was brought at one of my outreach OPDs (outpatient department) with 'seizures' since 4 years of age. The frequency of the episode was about once in six months. These events never occurred out of sleep. He would usually fall, become stiff, his lips would turn blue, he may feel 'sweaty' to touch and this would last less than two minutes. The child reported that he experienced 'blurring of vision' and could feel his heartbeats just before he fell. He was youngest of three offspring and the oldest one had also received treatment for 'seizure'. He was born of non-consanguineous marriage, his developmental milestones were normal for age and there was no history of neonatal admission.

He had a normal head circumference or OFC (occipitofrontal circumference), had hypertelorism, was right handed and did not carry any neurocutaneous markers.

His parents produced two EEG (electro encephalograph) studies that were reported as normal and MRI (magnetic resonance imaging) of the brain which was also reported to have no abnormalities. He was taking two antiepileptic drugs but the frequency of events remained unchanged.

This I thought, did not 'sound' like seizure, in fact it did not 'sound' like epilepsy!

I was convinced that these episodes were syncopal attacks.

Syncope is a transient and abrupt loss of consciousness with complete return to preexisting neurologic function. It is classified as neurally mediated (i.e., carotid sinus hypersensitivity, situational, or vasovagal), cardiac, orthostatic, or neurogenic. Vasovagal syncope (vay-zoh-VAY-gul SING-kuh-pee) is not uncommon in children and have an easily identified triggering event such as pain, sight of blood, prolonged standing and likewise.

In this case, the events were more than few and they were all in awake state, in upright position, but it was difficult for the parents to recall the trigger for each episode.

At the end of the consultation, I had put down the diagnosis as Long-QT syndrome (LQTS)

Further course

I sent the child for an ECG (electrocardiography) and waited with bated breath (and a working calculator)!

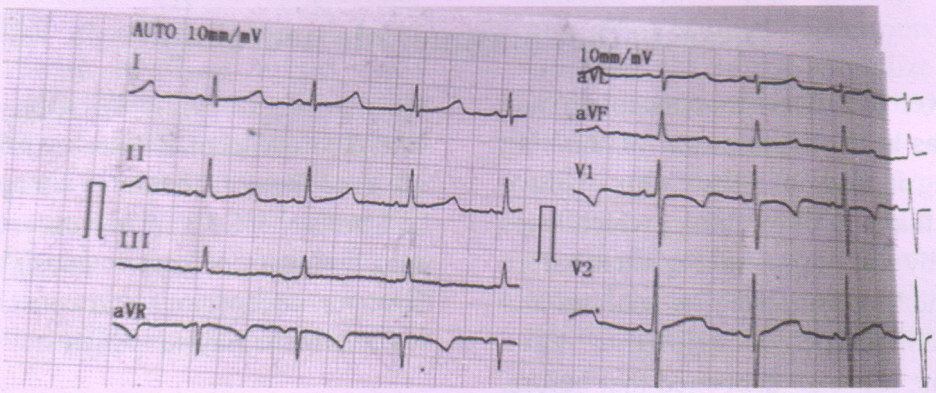
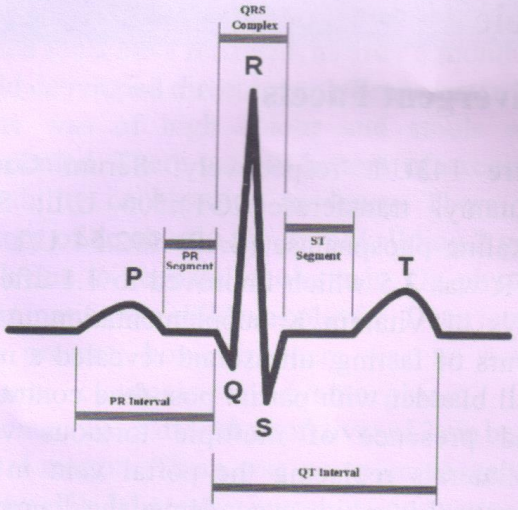


Figure 1: Standard 12 lead ECG of the patient on first visit



QTc = QT / √RR,
Where:
QTc = corrected QT,
QT = duration of QT interval,
RR = duration of RR interval.
(Bazett's formula for calculating QTc has its limitations – it is known for overcorrecting at high heart rates and undercorrecting at low heart rates.)

QTc= 9.5*0.04 divided by 18*0.04 =0.452 seconds

Although the duration of QT was prolonged, my analytical brain kicked in to cause confusion. A duration more than 480 milliseconds (ms) has a score of 3 in diagnosis of LQTS while in this case, QTc was 452 ms which had a score of 1. As per the LQTS diagnostic criteria, syncope with stress had a score of 2; making a total score of 3 which made an intermediate probability of LQTS.

I asked the parents to continue the antiepileptics and told them to follow up in a month's time when I visit the outreach clinic next.

To my immense good fortune, the parents brought the child for follow up the very next month. He had another event during this interval; he was playing and felt uneasy, as he was about to leave for home from playground, he fell down, lost consciousness and was brought home by his friends. His parents thought he was pale and dull when they received him. He had regained composure in the next few minutes.

I cursed myself in my head and told the parents that they needed to see an Electrophysiologist/ Cardiologist at the earliest, never mind the distance of 300 kms that they needed to cover to do so.

I was pleasantly surprised to receive a call from the electrophysiologist, on the very next day. In a matter-of-fact manner, he told me that the child had long QT syndrome. His ECHO (Echocardiography) Colour Doppler study was normal and a repeat ECG had revealed a longer QTc with T-wave alternans.

Two months later, I came to know that the child harboured a homozygous mutation at exon 5 of KCNQ1 (potassium voltage gated channel) gene. He was not doing well on beta adrenergic blocking drugs; he would probably require a left cardiac sympathetic denervation or an Implantable Cardioverter-Defibrillator.

My snap judgement was right but I doubted it. Many a times in medicine, as in life, we are right the very first time and then we mess it up by overthinking or analysing unnecessarily. I thanked the Almighty that due to the vigilance of the parents, I was saved from making a grave error and the child would receive adequate treatment.

Dr. Sarbani Raha
Pediatric Neurophysician
drsarbaniraha@rediffmail.com