



BMH Med. J. 2022;9(2):48-49. **Case Report**

A Case of Malignant Hyperthermia With a Genetic Diagnosis

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Introduction

Malignant hyperthermia is a hypermetabolic crisis caused by disordered calcium metabolism when the susceptible individual is exposed to the triggering agent. It is caused by a pathogenic mutation of the RYR1 gene. Here we report our experience with a suspected case of malignant hyperthermia which was confirmed posthumously with a genetic diagnosis.

Case report

A teenage boy who had sustained a subdural hematoma and facial trauma was posted for suturing of the facial lacerations. Under standard monitoring, general anaesthesia was administered with Inj. Fentanyl 100 microgram, Inj. Propofol 100 mg and intubated under Inj. Succinylcholine 100 mg. He was maintained on oxygen, nitrous oxide and isoflurane. Post intubation patient remained stable for half an hour, when his end tidal carbon dioxide (ETCO₂) started increasing gradually reaching a value of 45 mmHg. Minute ventilation was increased, but ETCO₂ kept rising. As we made attempts to take blood sample for performing acid blood gas (ABG) analysis it was noticed that the patient was febrile. A provisional diagnosis of malignant hyperthermia was made and management measures were initiated. Surgeon was informed and surgery was abandoned. Inhalational agent was discontinued and soda lime canister was changed. A new flushed anaesthesia workstation was made available. Femoral artery was cannulated for blood pressure monitoring and internal jugular vein was catheterized. Ice packs were applied. Sodium bicarbonate, insulin dextrose and propofol infusions were started. Dantrolene was not available at our institution or any other hospital in our city. By one hour post induction, his ETCO₂ was 135 mmHg, heart rate 140/min, blood pressure was 136/74 mm Hg and temperature was 40 degrees Centigrade. ABG analysis showed a pH 6.3, PaCO₂ 150 mm Hg and potassium to be 8.1 mEq/l. Two hours after induction he developed asystole which was revived with one cycle of cardiopulmonary resuscitation and he was shifted to intensive care unit. He continued to have multiple episodes of cardiac arrests and succumbed soon after. His blood sample was sent for genetic analysis and he had a heterozygous pathogenic mutation, c.1024G>C (p.Glu342Gln) in Exon 11 in the RYR1 gene.

His family was counseled to undergo genetic testing which has not been done yet.

Discussion

Malignant hyperthermia (MH) is a rare pharmacogenetic disorder which causes a hyper metabolic reaction due to disordered skeletal muscle contraction on exposure to triggering agents. The most consistent clinical features of an MH reaction include a rise in ETCO_2 , unexplained tachycardia, muscle rigidity, hyperthermia and acidosis.[1] Larach et al.[2] developed a scoring system to help diagnose MH and our patient had a score of 111 which gives an almost certain diagnosis of MH.

The ryanodine receptor 1 (RYR1) gene is the main gene implicated in MH. Molecular testing alone cannot be used to identify at risk individuals and In Vitro Contracture test (IVCT) is the gold standard for detecting MH susceptible individuals. Genetic screening can only be considered as an adjunct to IVCT in patients with a family history of MH.[3] There are reports of MH susceptible patients being anaesthetized uneventfully by avoiding the known triggering agents.[4]

Thought to be non-existent in our population till 2007, few cases have been reported till date.[5,6] MH can have varying presentations from florid to more subtle presentations which are not investigated adequately and are left unreported. There is an ethical question that remains when we take up patients for anaesthesia without keeping dantrolene available knowing MH can happen any time. Increased awareness and vigilance leading to early detection and treatment are the best options available presently. Since we do not have registries for reporting these incidents, case reports can serve as reminders and improve awareness on the incidence of MH in Indian population.

Conclusion

The emerging case reports of malignant hyperthermia in our population contradict the prior belief that malignant hyperthermia is non-existent in our patients. This fact underlines the importance of stocking dantrolene. High vigilance in detecting and removing triggering agents can help prevent the progression of MH.

References

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The consent for publication was obtained from the family of the deceased patient with the guarantee that their anonymity will be maintained.