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## CLINICAL VIGNETTE

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# A 50-Year-Old Male with Inherited and Acquired Pseudocholinesterase Deficiency

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### *Introduction*

Succinylcholine is a depolarizing neuromuscular blocking drug used to facilitate ideal intubating conditions during laryngoscopy. "Pseudocholinesterase, also known as butyrylcholinesterase, is a hepatically produced enzyme that metabolizes succinylcholine, in addition to ester local anesthetics like cocaine."<sup>1</sup> When abnormal function of pseudocholinesterase is present, metabolism and the return of normal neuromuscular function can be delayed by up to four to six hours. "Pseudocholinesterase deficiency can be both inherited in an autosomal recessive pattern as well as acquired in certain disease states such as liver disease."<sup>2</sup> We present a case of a patient with severe liver disease who had a prolonged response to succinylcholine and laboratory testing showing both quantitative and qualitative pseudocholinesterase deficiency.

### *Case Report*

The patient was a 59-year-old man seen for emergent Esophagogastroduodenoscopy (EGD) for active hematemesis. He was admitted to the hospital three days prior for rapidly progressing abdominal distension and lower extremity edema over the prior 2 weeks. The patient had a 35-year history of alcohol abuse/dependence, however had abstained from alcohol for the prior two months after noticing muscle wasting, fatigue and darker stools which he had attributed to alcohol use.

On admission his MELD-Na score was 33 with 53% estimated 3-month mortality, and he met the criteria for Child-Pugh Class C cirrhosis. He was anemic with hemoglobin 5.6 g/dl, his INR was 1.9, with Na 123 mmol/L, and Creatinine 2.75 mg/dl. Over the first three hospital days he received several transfusions of PRBCs, underwent paracentesis with 9L total volume removed, with improvement in Hemoglobin, creatinine and Sodium. However, on hospital day 4, he had large volume hematemesis and scheduled for emergent EGD evaluation. His hemoglobin was 6.6g/dl, his INR was 2.55 and he was hypotensive and tachycardic due to his hypovolemia. The ICU service started transfusion of PRBCs and the patient was sent for emergency endoscopy.

Given active hematemesis with high risk of aspiration, he had general endotracheal anesthesia with rapid sequence intubation using succinylcholine to secure the airway in the most expeditious fashion. The 107 kg patient was intubated using etomidate 30 mg and succinylcholine 120 mg, and also received 100mcg

of fentanyl and 60 mg of lidocaine at the time of induction. Anesthesia was maintained with inhaled Sevoflurane 1.5-2.5% , with 100% oxygen. The only other agent he received was ondansetron 4mg to prevent postop nausea and vomiting. The endoscopy showed two large and deep duodenal ulcers not amenable to endoscopic intervention with a large amount of residual blood in the gastric fundus and grade C erosive esophagitis. The patient remained hemodynamically stable throughout the procedure, and no issues were noted until after the procedure was completed after 7 minutes total EGD time.

Emergence from anesthesia was started, however after approximately 20 minutes there were no signs of spontaneous ventilation. No evidence of neuromuscular recovery was noted, with nerve stimulation four ratio 0/4 and the post tetanic count 0. After another 15 minutes in the endoscopy suite, there were still no signs of spontaneous neuromuscular recovery.

Given the low dose of rapid acting anesthetic agents given during the procedure, and the information from the nerve stimulator showing a lack of neuromuscular recovery a provisional diagnosis of pseudocholinesterase deficiency was made. He was taken to PACU intubated and sedated with a propofol infusion. To confirm the provisional diagnosis, laboratory studies for the Dibucaine number and Pseudocholinesterase serum levels were obtained. The patient was supported in the recovery room for approximately 4 hours, before beginning to display signs of spontaneous neuromuscular recovery. His sedation was weaned and after 30 additional minutes he was fully awake, alert and able to follow commands. He was extubated with spontaneous breathing trial without any further issues related to neuromuscular blockade.

The pseudocholinesterase laboratory studies returned 10 days after the episode and confirmed a quantitative pseudocholinesterase deficiency (257 IU/L, nl 3342-7586) as well as a possible abnormal phenotype of pseudocholinesterase associated with prolonged apnea. The dibucaine number was 66.9, which was within the reported ranges for Phenotype F and S2. Unfortunately, the patient died within a week after obtaining these results without available family to notify for further testing.

## Discussion

“Pseudocholinesterase deficiency (PCD) is a rare defect in the pseudocholinesterase enzyme normally produced by the liver.”<sup>3</sup> This enzyme plays a crucial role in the rapid hydrolysis of acetylcholine, various ester-based medications, and common anesthesia drugs such as succinylcholine, mivacurium, and ester local anesthetics. “Deficiency in this enzyme can have substantial anesthetic implications such as prolonged paralysis due to the delayed metabolism of these medications.”<sup>1</sup> This deficiency has both inherited and acquired forms. “The inherited form passes genetically in an autosomal recessive pattern with distinct phenotypes leading to varying degrees of enzymatic dysfunction.”<sup>4</sup> “Heterozygotes carry only one gene coding for the abnormal enzyme, while homozygotes possess both genes coding for the defective pseudocholinesterase enzyme.”<sup>5</sup> Resulting phenotypes, such as A, AS, U, and UA, exhibit different levels of enzymatic function. Acquired pseudocholinesterase deficiency can be noted in various disease states such as malnutrition, pregnancy, burns, liver disease, kidney disease, hemodialysis, myocardial infarction (MI), congestive heart failure (CHF), malignancy, and chronic infections all of which can decrease production. Furthermore, certain medications like steroids and cytotoxic agents can contribute to reduced enzyme levels.

“The dibucaine number is a valuable laboratory result used to assess the level of pseudocholinesterase deficiency.”<sup>5</sup> Normal pseudocholinesterase is inhibited by dibucaine, and the degree of inhibition is expressed as a percent. The percent inhibition may range from 0 to 100. Lower inhibition indicates significant impairment in the enzymes function and indicates higher susceptibility to the effects of medications whose metabolism is dependent on this enzyme. The dibucaine number serves as a diagnostic tool to differentiate between various phenotypes of pseudocholinesterase deficiency and to estimate the extent of enzyme dysfunction. “Various phenotypes exist (such as U, A, AS S1, S2, F), and each are associates with variable levels of enzymatic deficiencies.”<sup>4</sup>

Our patient illustrates how presentation of pseudocholinesterase deficiency can be further complicated by having a combination of acquired and inherited forms of this deficiency. Our patient’s dibucaine number of 66.9, is consistent with an abnormal phenotype S2 or F. However, his extensive liver disease, with MELD score of 33 likely contributed to acquired deficiency as well. The combination of an abnormal phenotype and decreased production can lead to profoundly decreased enzymatic function and prolonged paralysis, as seen in this patient. This patient could also have received exogenous pseudocholinesterase enzyme from the intraoperative blood transfusion. This case illustrates the multifactorial nature of pseudocholinesterase deficiency with complex presentations. Complexity can result in variable and unpredictable muscular paralysis in patients undergoing anesthesia. This unpredictability emphasizes the critical role of aggressive nerve monitoring during anesthesia administration. Moreover, intraoperative “monitoring of neuromuscular function can aid in

promptly detecting and addressing unexpected prolonged paralysis.”<sup>6</sup>

Intraoperative management and “treatment of suspected pseudocholinesterase deficiency first involves developing a differential diagnosis.” This includes considering “narcotic overdose, residual neuromuscular blockade, cholinergic crisis, myasthenia gravis, myasthenic syndrome, hypermagnesemia, hypophosphatemia, and hypokalemia.”<sup>6</sup> PCD diagnosis should be considered after ruling out these reversible causes. The patient should remain sedated and mechanically ventilated. This ensures patient comfort and safety while waiting for paralysis to resolve. Monitoring muscle recovery using a quantitative or qualitative nerve monitor is essential. “It is important to note that pseudocholinesterase deficiency can result in a phase II block.”<sup>3</sup> Thus, agents like neostigmine should be avoided as they can worsen the paralysis. It is prudent to simply allow that paralysis to wear off while the patient remains comfortable while sedated and mechanically ventilated. Once paralysis resolves, further evaluation is recommended. Testing dibucaine levels and activity confirms PCD. If positive, genetic testing should be offered to patient and relatives to determine carrier status.

## Conclusion

Pseudocholinesterase deficiency poses unique challenges in anesthesia. This challenge is exacerbated by the multifactorial etiology causing the deficiency of this enzyme as illustrated in our patient. A comprehensive approach is required to evaluate a suspected pseudocholinesterase deficient patient. This involves ruling out other reversible causes from the differential diagnosis, followed by appropriate supportive care until the acute paralysis is fully resolved. Ultimately management must be followed up with appropriate laboratory testing and if warranted, genetic analysis to further elucidate the cause plan for future safe anesthesia.

## REFERENCES

1. **Andersson ML, Møller AM, Wildgaard K.** Butyrylcholinesterase deficiency and its clinical importance in anaesthesia: a systematic review. *Anaesthesia*. 2019 Apr;74(4):518-528. doi: 10.1111/anae.14545. Epub 2019 Jan 1. PMID: 30600548.
2. **Faber M.** Serum cholinesterase in disease. *Acta Medica Scandinavia*. 1943;114:59–71.
3. **Trujillo R, West WP.** Pseudocholinesterase Deficiency. 2023 Jul 10. In: *StatPearls* [Internet]. Treasure Island (FL): StatPearls Publishing; 2023 Jan–. PMID: 31082076.
4. **Jasiecki J, Jońca J, Żuk M, Szczoczarz A, Janaszak-Jasiecka A, Lewandowski K, Waleron K, Wasąg B.** Activity and polymorphisms of butyrylcholinesterase in a Polish population. *Chem Biol Interact*. 2016 Nov 25;259(Pt B):70-77. doi: 10.1016/j.cbi.2016.04.030. Epub 2016 Apr 22. PMID: 27109752.

5. **Rosenman KD, Guss PS.** Prevalence of congenital deficiency in serum cholinesterase. *Arch Environ Health*. 1997 Jan-Feb;52(1):42-4. doi: 10.1080/00039899709603798. PMID: 9039856.
6. **Robles A, Michael M, McCallum R.** Pseudocholinesterase Deficiency: What the Proceduralist Needs to Know. *Am J Med Sci*. 2019 Mar;357(3):263-267. doi: 10.1016/j.amjms.2018.11.002. Epub 2018 Nov 10. PMID: 30578021.