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CLINICAL QUIZ

Dangerous hyperkalemia in a newborn: Questions

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Clinical presentation

An 8-day-old male baby presented to our emergency unit with lethargy and poor feeding and reduced urine output since 3 days. He was a second born child of a third degree consanguineous marriage, born at term with a birth weight of 1.8 kg with no history of significant perinatal events. However, a history of sibling death following a similar illness at day 15 of life was reported.

On examination, the child was dehydrated, the skin was mottled, and peripheral pulses were weak. The child was in shock and had a convulsion while in the emergency department. He received saline boluses and supportive management was instituted, while his investigations were awaited. He had severe respiratory distress due to pneumonia for which he required mechanical ventilation and inotropes were started for shock. IV antibiotics were started in view of positive screen for sepsis. His serum potassium was 11.9 mEq/dl at admission and serum sodium was 119 mEq/dl. Blood urea was 78.42 mg/dl and serum creatinine was 1.41 mg/dl initially, which was subsequently documented normal (23 mg/dl, 0.4 mg/dl respectively). In view of severe hyperkalemia, he was started on peritoneal dialysis (PD) along with supportive management with sodium bicarbonate, salbutamol nebulization and potassium binding resins through nasogastric tube.

The baby improved after supportive management for sepsis and was off inotropes after 48 h of admission. His potassium improved after 12 h of PD ($K^+ = 3.7$ mEq/dl) after which PD was stopped. Forty-eight hours later, the child was weaned off the ventilator but he again developed hyperkalemia (serum potassium of 9.6 meq/dl) and hyponatremia (serum sodium of 114 mEq/dl) and PD had to be instituted again. The child developed a skin rash (miliaria rubra) the following day which improved a week later and during his prolonged course of stay in the hospital he developed pneumonia, which again required him to be ventilated for 7 days. During his hospital stay, the child required a total of 8 PD sessions, each time owing to uncontrollable hyperkalemia.

Questions

1. What is the differential diagnosis of such a presentation in a neonate?
2. How would you confirm the diagnosis?
3. What is the genetic/molecular basis of such a disorder?
4. What is the optimal management?

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Compliance with ethical standards

Conflict of interest The authors declare that they have no conflict of interest.

The answers to these questions can be found at https://doi.org/10.1007/s00467-018-4102-4.

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