Convulsive satus epilepticus with neurocutaneous syndrome

Title: Management of case of convulsive status epilepticus with neurocutaneous syndrome

Introduction

Status epilepticus is life-threatening neurological disorder defined as 5minutes or more of continuous seizure without complete recovery of consciousness between seizures. It has two foms; Generalized convulsive SE & Non-convulsive SE. Convulsive SE is more common in children. Commonly caused by fever, genetic predisposition, metabolic and electrolyte disturbance or head trauma, CNS infection.

Neurofibromatosis is a common autosomal dominant genetic disorder. Classified into two types: (NF1) which is more common in children and (NF2) which manifest later in adulthood. It is neurocutaneous syndrome characterized by diagnostic skin lesion called café au lait patches and neural manifestations which may not be obvious and only presented when complicated by seizures which is considered an emergency case and need immediate management.

Methods We searched several internet databases as Pubmed, Cochrane, Medscape, Science Direct Trip databases , and the Journal of American Academy of Neurology. Meta analysis, clinical trials, systematic reviews and randomized control trials and other observational studies concerning Status epilepticus, its investigations and different anti-epileptic drugs. In addition neurofibromatosis and Tuberous sclerosis were reviewed, as well

Aim to know how to diagnose, treat & follow up this case to prevent further remission or complications

Result

We came across 280 research and case report describing status epilepticus and its management. Status epilepticus is an emergency situation that needs rapid management, but if refractory and continues more than 60-90 minutes after initiation of therapy mortalities increase. Status epilepticus associated with cafe au lait patches is characterstic to neurocutaneous syndrome most probably neurofibromatosis type1. Status epilepticus may be complicated by Acidosis, Respiratory distress, Fever which is mediated by extreme muscle activity rather than infection, initial release of catecholamines into circulation causing increase in blood pressure and heart rate causing cardiac arrhythmia, Catecholamine excess causing hyperglycemia[1]. Saving life by maintaining ABC, Introducing vascular access and giving lorazepam are the first line in management, If there is no response within 10 minutes give another dose of lorazepam, If there is no response within 10 minutes give phenytoin, If convulsions persist more than 20 minutes use general anesthesia and put the patient on mechanical ventilation. Buccal midazolam is as effective as rectal diazepam but we can use rectaldiazepamif preferred or if buccal midazolam is not available. No evidence supporting dexamethazone in status epilepticus treatment [2]. No evidence supporting the use of antibiotics in case of seizures caused by Nf1, the only indication to use antibiotics if meningitis is the cause of seizures[3]. Asking for history of anticonvulsant and drug levels assesment is important as there is significant variation in effect and response to anti-epileptic drugs as seizures may

develop even with taking themas; in case of low dose of anti-epileptic drug, non-compliance to antiepileptic drugs. Adding new antiepileptic drugs to old ones to decrease seizures is required in some cases[4]. No enough evidence supporting blood transfusion in status epilepticus treatment. No evidence supporting direct effect of status epilepticus on platelet count except in rare condition of lorazepam toxicity that leads to abnormally decreased platelet count. Mitochondrial encephalopathy, Lactic Acidosis and Stroke-Like Episode presents in pediatric age group so pyruvate and lactate assessment is important to detect the cause and severity of status epilepticus[5]. Blood sugar Assessment is important as duration & extent of glucose dysregulation could be predictor of pathological outcome of status epilepticus as seizures can be exacerbated in cases of hyper or hypoglycemia and become resistant to antiepileptics when blood glucose is not controlled[6]. Electrolyte assessment including (Na, ca and cl) is important as seizures result from loss of balance between intracellular and extracellular chloride concentration causing hyper-excitability and Neuronal death result from large increase in intracellular calcium[7]. Genetic testing involving exome sequencing may help in reaching accurate diagnosis and may reveal novel autosomal recessive genes associated with idiopathic epilepsy. No enough evidence supporting abdominal sonar[8]. Brain Imaging can detect the etiology of status epilepticus espicially MRI, It helps to identify and localize epileptic foci[9]. CSF analysis is important in detecting cause of status epilepticus as it may diagnose autoimmune disorder when there is high titre of CSF GABAA receptor antibodies or showing in vivo biomarkers for neuronal damage after epileptic seizure10].

Conclusion

This child is suffering from Status epilepticus which is most probably caused by neurofibromatosis type1. For investigation, I recommend immediate assessment of blood sugar level, electrolytes disturbance, purvate and lactate levels and antiepileptic drug level (in case of history of intake). Later on after stabilization of condition, I recommend brain imaging, cerebral fluid analysis and genetic testing. He is prone to be complicated by acidosis, respiratory distress, fever, increase in blood pressure and heart rate, arrhythmia and hyperglycemia. Status epilepticus with NF1 is very serious due to its high mortality rate. Modulating the dose and the type of antiepileptics is a must according to the drug level and history of intake. Lorazepam intravenous is better than rectal diazepam in this situation. There is no evidence recommend dexamethasone, antibiotics, blood or platelets transfusion. reference Pollard H, Cantagrel S, Charriaut-Marlangue C, Moreau J, Ben Ari Y. Apoptosis associated DNA fragmentation in epileptic brain damage. Neuroreport 1994; 5: 1053-5. [1]

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