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Sulphite Oxidase Deficiency: A Rare Case Report

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Abstract **Case Report**

Sulphite oxidase deficiency is known to be a rare autosomal neurometabolic disorder in turn which is characterized by neonatal-onset encephalopathy mimicking hypoxic-ischemic insult with intractable seizure. The report shows a three years old girl presenting with intermittent ataxia, recurrent seizures and uncoordinated body movements and bilateral lens dislocation. Further investigations led to confirmation of isolated sulfite oxidase deficiency. This case represent the clinical variability of SOD and it is not only atypical but also seems to be the mildest form described so far. The association of ectopia lentis with intellectual disability make us to look for this diagnosis.
Keywords: Sulphite oxidase deficiency.

INTRDUCTION

Sulphite oxidase deficiency is known to be a rare autosomal disorder of the nervous system in new born. Sulfite oxidase is a mitochondrial enzyme coded by the gene SUOX and mutation in this gene cause impair the function of sulfite oxidase, preventing complete breakdown of sulfur-containing amino acids. As a result, there is excessive accumulation of Sulphite & S-sulphocysteine in body, which is toxic for nervous system that results in the brain damage [1, 2]. Other factor which can cause deficiency of sulphite oxidase is defects in molybdenum cofactor synthesis. Molybdenum cofactor deficiency also leads to deficiency of xanthine dehydrogenase and aldehyde oxidase [3]. Sulphite & S-sulphocysteine can be detected through urine and plasma of the patients; however Serum uric acid and urinary excretion of xanthine and hypoxanthine are normal in sulphite oxidase deficiency individual [4]. The initial symptom of all the patient was seizures, which was reported to be the most common presenting symptom of this disorder [5]. Sulphite oxidase deficiency is inborn error which is characterized by dislocation of ocular lenses, mental retardation, and in severe cases, attenuated growth of the brain[6].

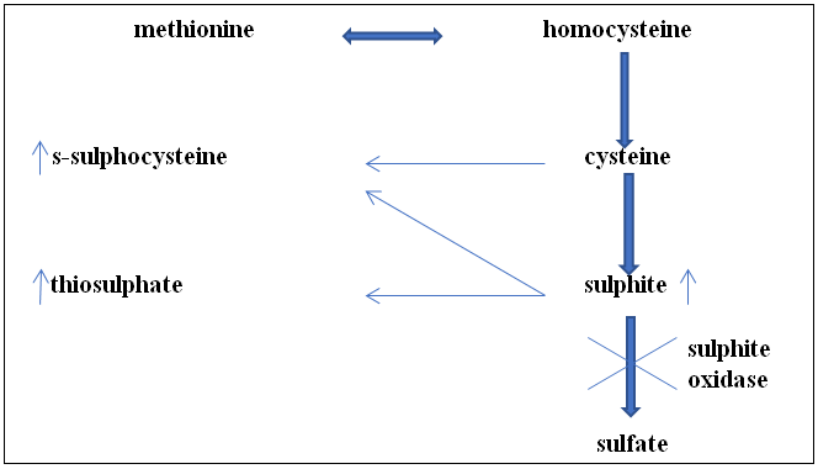

Fig-1: Metabolic pathway of sulfur containing amino acid and role of enzyme sulphite oxidase

CASE REPORT

A 3-year girl was reported to us for the biochemical analysis with complaints of ataxia, recurrent seizures, weakness, palpitation, excessive sweating, giddiness and vomiting since many days. She also has learning disabilities. Her height and weight were 102 cm and 19 kg respectively. The following biochemical examination of urine and plasma was made to identify sulphite oxidase deficiency, increased level of SGOT, SGPT & creatinine was observed in plasma. At initial assessment, she was obtunded and showed severe neurological impairment whereas dystonia, recurrent opisthotonic posturing had been observed.

Serum and urine uric acid levels were normal, excluding molybdenum cofactor deficiency. Urinary xanthine and hypoxanthine levels were normal. These laboratory findings of normal serum and urine uric acid, xanthine, hypoxanthine; however, a positive urine sulfite dipstick test, elevated urinary S-sulfocysteine and low plasma cysteine with raised plasma taurine were identified with the diagnosis of isolated sulfite oxidase deficiency. The excretion of urinary sulfocysteine seems to be very high 148 umol/g crt (ref. value: < 66 umol/g crt). Whereas the plasma cysteine level was decreased 10 mol/L (ref. value: 18-122 mol/L).

Electroencephalography was performed observe asynchronous burst suppression tends to early infantile epileptic encephalopathy. Epileptic encephalopathy is assumed due to the accumulation of toxic metabolites due to deficiency of enzyme sulphite oxidase. Brain computerized tomography reveals diffuse cerebral edema shows an ischemic insult. Other investigation with normal ammonia, serum bicarbonate, lactate. She died after the one weak of hospital discharge.

DISCUSSION

Sulphite oxidase is a hereditary metabolic disorder and autosomal recessive trait. It is rare case few cases have been reported all around. It is an enzyme in the mitochondria of all eukaryotes. It oxidizes sulfite to sulfate via cytochrome C and transfers the electrons produced to the electron transport chain, allowing generation of ATP in oxidative phosphorylation and deficiency leads to altered ATP production which affect the entire metabolism processes [7]. In mammals, the expression levels of sulfite oxidase are high in the liver, kidney and heart, and very low in spleen, brain, skeletal muscle, and blood [7]. It affects the brain cell causing encephalopathy and it is over all due to accumulation of toxic metabolites which are sulphite and are strong nucleophile which in turn react with protein disulphide to produce sulphonated cysteine derivatives. Hence, alteration of protein structure by sulfitolysis result various biochemical changes in the body [6].

CONCLUSION

To date, no effective rehabilitation is available for sulphite oxidase deficiency, and death in early stages has been the usual consequence. So further more research is required for early diagnosis & treatment.

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