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Letter to the editor

Neuropathological findings in a case of combined deficiency of sulphite oxidase and xanthine dehydrogenase

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Sir, with interest we have read the recent paper by *Roth et al. 1985* [3] in this journal on *neuropathological findings in a case of inherited combined sulphite oxidase and xanthine dehydrogenase deficiency*. This abnormality is due to the absence of molybdenum cofactor, an essential constituent of both enzymes [2,5]. To date nine cases are known [1]. As far as we know four patients are presently surviving.

We wish to report on neuropathological observations in a similar patient. The proband M.V. was born as the second child of healthy non-consanguineous parents. More detailed data on this case have been reported elsewhere [5]. Severe seizures started soon after birth. He died at the age of eight days, despite intensive treatment. The diagnosis combined deficiency of sulphite oxidase and xanthine dehydrogenase could be confirmed in an autopsy liver specimen.

Histopathological findings in the brain included shrinkage and severe loss of neurons, particularly in all layers of the isocortex, thalamus and basal ganglia. Paucity of myelin in the long tracts was noticed with isomorphic gliosis.

The most interesting finding was a diffuse spongiosis, affecting both the neuropil and white matter structures (Fig. 1).

The findings in this brain bear an obvious similarity to those in isolated sulphite oxidase deficiency as reported by Rosenblum [4] and in the combined deficiency by Roth et al. [3]. As stated by the latter, the neuropathological findings are not specific.

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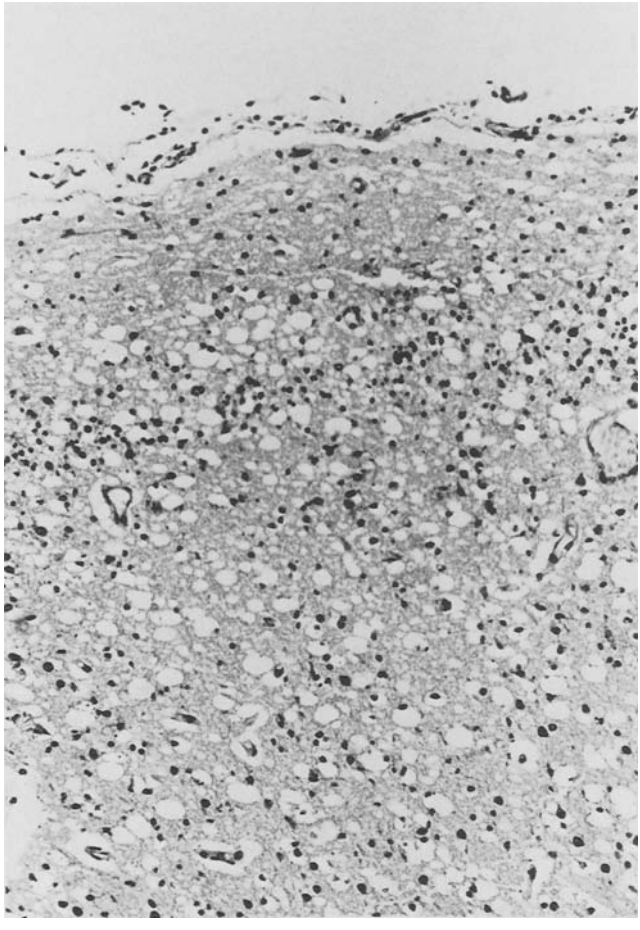

Fig. 1. Neocortex of patient with severe loss of neurons and extensive spongiosis in all layers. HE × 132

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