LETTER TO THE EDITOR

Hereditary persistence of alpha-fetoprotein

Dear Editor,

The article by Houwert *et al*. about Hereditary persistence of alpha-fetoprotein (AFP)[1] correctly underlines that increased serum AFP concentrations do not necessarily reflect malignant or hepatic disease. Unfortunately, their list of conditions (*table 4*) associated with increased serum AFP is incomplete.

Ataxia with oculumotor apraxia type 2 (AOA2), ataxia telangiectasia (AT) and variant AT are three conditions belonging to the autosomal recessive ataxias that are characterised by increased serum AFP levels.[2] For (variant) AT, AFP may be in de range of 200 to 500 ng/ml.[3] All three conditions are caused by a defective DNA repair mechanism, which is most seriously affected in AT. Particularly in AT and variant AT, the diagnosis should not be overlooked. First, because various complications may develop including diabetes mellitus, immunodeficiciency and severe lung restriction. Second because maligancy often develops in the 2nd or 3rd decade. Third, because the increased radiosensitivity precludes the use X-ray diagnostics, radiotherapy and alkylating agents.

We feel the need to emphasise the presence of high serum AFP levels in AT, because the diagnosis can easily be missed for several years, particularly in patients with variant AT who may have only subtle neurological problems until adulthood. In these cases, high AFP levels may be the key to the correct diagnosis and appropriate patient management. Ignorance of the diagnosis of (variant) AT will put a patient at risk after mechanical ventilation,[4] when X-ray diagnostics are perfomed, or – in case of proven malignancy – when DNA-damaging therapy is given.

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