

## Re: Consider Muscle Disease in Children with Elevated Transaminase

*To the Editor:* We read with great interest the brief report by Wright et al,<sup>1</sup> which emphasizes that high levels of transaminases may be observed early in presymptomatic children with an underlying myopathy and can wrongly raise the suspicion of an underlying liver disease. According to the results of study by Wright et al, the majority of participants turned out to carry a pathogenic mutation in the dystrophin gene. On the basis of these observations, they proposed that measurement of serum creatine kinase should be obtained when clear gastrointestinal symptoms and signs are absent, and they stressed the need to combine the laboratory findings with a neurological examination.

This notion may prove to be even more useful in the work-up of asymptomatic or oligosymptomatic adults with a muscle disease who are found during a routine examination to have increased aspartate aminotransferase and alanine aminotransferase levels. It is interesting that there are patients with a metabolic myopathy caused by acid maltase deficiency who may present with mild increase in transaminases and a concomitant mild elevation of creatine kinase.<sup>2,3</sup> The correct diagnosis is imperative for prompt treatment, especially in this era of enzyme replacement therapy. Moreover, it provides the possibility of prenatal diagnosis and can prevent patients from submitting to unnecessary tests such as liver biopsy.

Another interesting point, according to the suggestions of Wright et al,<sup>1</sup> is that elevated serum  $\gamma$ -glutamyl transpeptidase can be used to identify patients with liver disease. However, it must be mentioned that  $\gamma$ -glutamyl transpeptidase frequently is elevated in patients with myotonic dystrophy type 1 (Steinert disease), which may indicate mild liver involvement, although there is no clinically significant liver disease.<sup>4,5</sup> So much depends on clinical examination, which may reveal the characteristic phenotype including hatchet face, blepharoptosis, and frontal balding with or without clinical myotonia.<sup>6</sup>

Therefore, on the grounds of these considerations, it is well concluded that no rigid laboratory rule can be laid down for distinguishing a myopathy from a pure liver disease, and laboratory markers must be evaluated cautiously to minimize any risk of misdiagnosis.

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