Dihydropyrimidine dehydrogenase deficiency caused by a novel genomic deletion c.505_513del of DPYD

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Dihydropyrimidine dehydrogenase (DPD) deficiency is an autosomal recessive disorder of the pyrimidine degradation pathway. In a patient presenting with convulsions, psychomotor retardation and Reye like syndrome, strongly elevated levels of uracil and thymine were detected in urine. No DPD activity could be detected in peripheral blood mononuclear cells. Analysis of the gene encoding DPD (DPYD) showed that the patient was homozygous for a novel c.505_513del (p.169_171del) mutation in exon 6 of DPYD.
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