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P-134 - TRANSIENT 5-OXOPROLINURIA (PYROGLUTAMIC ACIDURIA) INDUCED BY POLIMEDICATION COMBINED WITH ACETAMINOPHEN

Velasquez-Rivas D¹, Maccarone M¹, Fuertes A¹, Sokn S¹, Villanueva MM¹,², Dominguez R², Durand C¹, Massaro M², Schenone AB¹

(1) FESEN-Laboratorio de Neuroquímica “Dr. N.A. Chamoles”. (2) F.L.E.N.I. C.A.B.A.-Argentina. dvelasquez@laboratoriochamoles.com.ar

INTRODUCTION: pyroglutamic aciduria is an inborn error of metabolism due to the deficiency of glutathione syntetase or 5-oxoprolinase in the gamma-glutamyl cycle; it can also be detected secondary to the treatment with acetaminophen in conjunction with antibiotics or antiepileptic drugs. These drugs saturate the cycle and produce elevation of 5-oxoproline (pyroglutamic acid, PA) leading to metabolic acidosis. CASE: A 12 month-old (mo) female patient was on treatment with antibiotics and antiepileptic drugs because of sepsis and refractory seizures respectively. Multiple metabolic tests had been done since 2 mo and PA was detected in urine organic acids (UOA) for the first time at 12 mo. In a second sample we also found acetaminophen metabolites. The PA elevation was consistent with a deficiency in the gamma-glutamyl cycle but other clinical and biochemical features didn´t support the diagnosis. We asked for another sample to repeat UOA but requested to stop acetaminophen intake and obtained a completely normal result. Finally, the patient underwent chromosomal microarray testing resulting in a gross deletion of 9.7 Mb (2q24.3-q31.1) associated to complex epileptic syndromes. CONCLUSIONS: we assume the elevation of PA in our patient was secondary to the treatment with acetaminophen in combination with other drugs, because the discontinuations of acetaminophen normalized UOA. We emphasize the idea that clinical, pharmacological and nutritional information is essential for the interpretation of UAO.