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Biochemical Diagnosis of a Fatal Case of *Günther's* Disease in a Newborn with Hydrops Foetalis

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Summary: The birth of a male baby was induced at 32 weeks. In utero, the child presented, inter alia, signs of hydrops, hepatosplenomegaly and anaemia. Two in utero transfusions for correction of the anaemia were performed at 28 and 29 weeks, respectively. The baby rapidly presented respiratory distress with mixed acidosis. Three hours after birth, pink urine was excreted. Signs of icterus necessitated phototherapy, after which photosensitivity occurred. Erythrocytes were fluorescent under long-wavelength UV light. The baby died 24 hours after birth, displaying severe acidosis, a diffuse haemorrhagic syndrome, and repeated bradycardia which did not respond to isoprenaline.

The analysis of porphyrins in urine, blood and faeces of the baby gave the following results:

- 1) uroporphyrin (I and III isomeric series) was increased in urine and faeces, with traces in erythrocytes and plasma;
- 2) heptacarboxyporphyrin I was found mainly in urine and much less in erythrocytes, plasma and faeces;
- 3) coproporphyrin I was increased in urine, erythrocytes, plasma and faeces, and
- 4) 5-aminolaevulinic acid and porphobilinogen in urine and plasma were within the reference ranges.

Determination of the enzymes of haem biosynthesis in erythrocytes and lymphocytes showed that both parents possessed only 50% of the normal activity of cosynthase.

A previously described point mutation in codon 73 was observed in one parent. Fatal cases of neonatal *Günther's* disease are extremely rare and such an observation, according to our knowledge, is probably one of the first described.

Introduction

Günther's disease, also called congenital erythropoietic porphyria (CEP), is a very rare (less than 200 cases reported) type of porphyria (1, 2). Congenital

erythropoietic porphyria is characterized by an overproduction of porphyrins of isomer I series, with accumulation of these compounds in bone marrow erythroblasts, teeth and bones (1, 2). These porphyrins