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Congenital erythrocytic porphyria: A case report

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Congenital Erythrocytic Porphyria is a rare congenital disorder of heme synthesis inherited as auto soma recessive trait. Clinical manifestations can range from mild to severe. This condition can be missed in early life as hematologic presentation may predominate than dermatologic manifestation. This is a case report of an infant who presented with history of blood transfusions for anemia after 6th month of life, red urine and a rash on face. On investigations her work up came positive for porphyria putting the whole constellation of symptoms, signs and investigations the diagnosis of congenital erythrocytic porphyria was made. In conclusion, early recognition of this condition may prevent skin complications which tend to increase with sun exposure leading to disfigurement in children later in life.

Biography

Arshalooz J Rahman is a graduate of Dow Medical College Karachi, Pakistan. She has completed her Residency from Brookdale University Hospital in Pediatrics and Fellowship in Clinical Pediatric Nephrology from Stony Brook University Hospital, USA and Diploma in Health Professional Education from Aga Khan University. She is Diplomate American Board of Pediatrics and presently working at The Aga Khan University Hospital in the Department of Pediatrics and Child Health. Her clinical and academic interest is in general pediatrics, pediatric nephrology and medical education. She is the Training Supervisor for FCPS and MRCP candidates at her institute and Examiner for postgraduates at College of Physicians and Surgeons, Pakistan.

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