ABSTRACT BOOK ABSTRACTS



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GENETICS AND GENODERMATOSES

HOW DO WE EXPLAIN VITAMIN D RICKETS IN TUNISIAN PATIENTS WITH LAMELLAR ICHTHYOSIS IN A SUNNY COUNTRY?

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Background: Lamellar ichthyosis is a rare genodermatosis belonging to the spectrum of autosomal recessive congenital ichthyosis. Children with lamellar ichthyosis especially in children with pigmented skin phototype IV are with increasing risk to develop vitamin D deficiency and secondary rickets.

Observation: A 13-year-old Tunisian male child of phototype IV born as a collodion baby from a consanguineous marriage was referred to our department for a congenital lamellar ichthyosis. His dermatological examination revealed thick adherent lamellar scales covering his entire body. He presented with a history of progressive knee deformity and difficulty in walking of 3 years duration. His mother reported that because of social embarrassment, he avoided exposure to sunlight. On examination, he had bone deformities with a waddling gait. Biological investigations revealed low calcium levels 1.3 mmol/l, high level of alkalin phosphatase 650UI/l, low vitamin D level 3 UI/l. PTH levels were also elevated. Skeletal survey showed features of advanced rickets. In fact, radiological investigations of the hips and wrists demonstrated features of rickets in the form of splaying and fraying of the distal ends of the bones with loss of provisional zones of calcification. Based on his clinical, biological and radiological findings, the diagnosis of lamellar ichthyosis and rickets was diagnosed. The patient received vitamin D and calcium supplementation with a slight improvement of his bone deformities. Regular sun exposure and a diet rich in vitamin D and calcium were advised to our patient.

Conclusions: This case is reported to highlight the possible association of severe lamellar ichthyosis with vitamin D rickets. This complication is owing to alterations in epidermal cholesterol metabolism possibly involving vitamin D receptors. Increased keratinocyte proliferation may result in poor or no penetration of skin by sunlight even in a sunny country as in Tunisia.





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