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Global Diabetes 2021: Netherton Syndrome: A Family Case Study of Phenotypic Variance and Literature Review

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Netherton's syndrome is a rare autosomal recessive disorder caused by a germline mutation in the SPINK5 gene which encodes LEKTI. The purpose of LEKTI is to inhibit kalikreins which are epide4rmal proteases important for the optimization of the epithelial structure. It is often diagnosed in the neonatal period to individuals who present with congenital ichthyosiform erythroderma. These individuals will also show trichorrhexis ingaginata, severe atopy, and ichthyosis linearis circumflexa. The severity and variability of the phenotype is dependent on the location of the mutation within the SPINK5 gene. It is important for this disease to be identified and treated early as more timely treatment is associated with reduced mortality, especially in the neonate period. The lack of awareness of Netherton's syndrome often leads to misdiagnosis of atopic dermatitis or psoriasis. In this case we present two children with various phenotype and complications born to a mother who has a previously diagnosed child with Netherton'ssyndrome.

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