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CONGENITAL ICHTHYOSIFORM ERYTHRODERMA AND ALPERT-SCHONBERZ DISEASE

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Congenital Ichthyosiform Erythroderma (CIE) is an autosomal recessively inherited disease of keratinization and is characterized by scales and hyperkeratosis where as osteopetrosis (Alpert-Schonberz disease) is an inherited condition characterized by generalized increase in skeletal density. Coexistence of these diseases is rarely reported. A 5.5 year-old boy presented in our Department with lumbar and inguinal pain. Since birth, he has had scales, sparse hair and nail dystrophy and CIE was diagnosed. During clinical examination craniofacial disproportion was also noticed. Skeletal roentgenograms showed generalized increase in the density of iliac, sciatic, pubic bones, the lower lumbar vertebrae, the sacrum and the femur and loss of medullary cavity. The base and the calvaria of the skull were also dense and thickened. All laboratory tests except for the calcium metabolism, were normal. Visual function was also normal. He was put on a low calcium diet and he was suggested to take 1-OH- cholecalciferole. Prognosis was not considered good. Conclusions: The coexistence of these two conditions is extremely rare but possible. Specific genetic studies are needed to investigate any relationship between the two that could explain this case.