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ONE PATIENT WITH TWO RARE DISEASES – INDIVIDUAL APPROACH IN GROWTH HORMONE DEFICIENCY (GHD) TREATMENT WITH rGH IN COFFIN-SIRIS SYNDROME

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Coffin-Siris syndrome (CSS) is a genetic disease inherited in an autosomal dominant manner, but most reported cases are caused by mutation of AR1D1B gene, which occurs *de novo*. Its characteristic features are: developmental or cognitive delay of varying degree, face and body dysmorphy, fifth nail/distal phalanx hypoplasia/aplasia, hipertrichosis, height deficiency, malformations affecting the CNS and cardiac and genitourinary systems. Even thought short stature is characteristic for this syndrome, growth hormone (GH) levels are usually correct.

In order to diagnose of growth retardation, 13 years old patient with CSS has been admitted to the Department of Pediatrics, Endocrinology, Diabetology with Cardiology Divisions of the University Children's Clinical *Hospital* in Białystok. Patient has specific features like viscerocranium dysmorphy, motility disorder of gastrointestinal tract, corpus callosum agenesis, heart defect, developmental delays and intellectual disability. From the age of about five, a slowdown in growth was observed. When he was 13 years and five months old the growth track was significantly below the 3rd percentile. Radiological bone age was estimated on about 11 years. Hypercortisolemia and thyroid dysfunction were ruled out. Due to low growth hormone release in the nocturnal profile and stimulation tests, growth hormone deficiency (GHD) was diagnosed. Growth hormone treatment was administered, resulting in accelerated growth rate.

This case reveals us three significant findings. Firstly, rare diseases do not rule out coexistence of other conditions. Secondly - a patient with low growth and concomitant genetic syndrome should have a hormonal diagnosis. At least – GHD treatment in a patient with Coffin-Siris syndrome gives the child a chance to improve final height.