Hereditary causes of rickets often take longer to be diagnosed because they are not very frequent. 1α-hydroxylase alteration is the most frequent vitamin D-dependent rickets. Beginning of treatment associated to a late diagnosis generates an abrupt parathormone deficiency. When initiating treatment, these patients have a risk of hungry bone syndrome.

CASE

2-year-old patient, male. History of psychomotor retardation, cellular immunodeficiency, short stature, epilepsy, and suspicion of skeletal dysplasia. Patient is hospitalized in Intensive Care Unit due to pneumonia. Tests results showed ionic calcium 1.43 mg/dL, phosphorus 2.5 mg/dL, magnesium 1.3 mg/dL, alkaline phosphatases 963 U/L, parathormone 225 pg/mL, vitamin D 59.43 ng/ml, TRPi 70.48%, creatinine 0.15 mg/dL. X-rays present generalized osteopenia with widening and cupping of the metaphyseal regions and multiple long bones fractures. Vitamin D-dependent rickets is diagnosed and later confirmed with genetic study: CYP27B1 c.[196-2A>G]; [1226C<T]. When admitted, patient initiates treatment with calcitriol 0.25 ug every 12 hours with dose titration up to 5 ug every 12 hours, due to differential diagnosis of vitamin D receptor alteration, prior to genetic test result; increase of oral and intravenous calcium needs up to 500 mg/kg/day of elemental calcium, alongside magnesium and phosphorus.

With lowered parathormone and alkaline phosphatases the first 10 days, but then an increase up to 300 pg/ml and 1250 U/L respectively with normal levels on day 35. After two months of follow up, patient is supplemented with low doses of calcium and calcitriol 0.5 ug every 12 hours and evolves with progressive catch up in size, recovers developmental milestones progressively, resolves immunological efficiency and convulsive syndrome.

CONCLUSIONS

Rickets is a differential diagnosis that we should not leave aside. Even though it is less frequent, genetic causes exist. Not only endocrinologists should be familiar with it, but also pediatricians, neurologists, immunologists, geneticists, and pulmonologists due to its systemic repercussion. The beginning of treatment in long-term diseases has bone remodeling level consequences similar to patients post parathyroidectomy in hyperparathyroidism, which leads to bone remodeling unbalance due to cessation of resorption stimulus, which is why it is important to early detect hungry bone syndrome: hypocalcemia, hypophosphatemia, hypomagnesemia and give prompt treatment.