Joubert syndrome coexisting with combined pituitary hormone deficiency in infant boy - case report.

Authors: ALICJA PIETERSZEK, Karolina Rutkows, Anna Wędrychowicz

Institution: Jagiellonian University Medical College, Students’ Scientific Group at Department of Pediatric and Adolescent Endocrinology, Cracow, Poland

Introduction: Joubert syndrome is a genetic disorder that belongs to ciliopathy group. It is characterized by brain abnormalities, hypotonia leading to ataxia and delayed development. Prevalence of the disease is 1:80,000, but it may be underestimated due to heterogeneous manifestation. Combined pituitary hormone deficiency is a dysfunction of at least two of pituitary-organ hormonal axis. It affects 1 out of 100,000 children. Signs and symptoms depend on child age and in infant period include seizures, prolonged jaundice, hypotonia, micropenis and delayed development.

Case presentation: In the thesis there is presented a case of an infant boy with multiple abnormalities in physical examination as well as general clinical picture. After delivery he presented with seizures, multisystem organ failure and improper sucking reflex. On physical exam he presented with asymmetric legs and ears setting, micropenis, hypotonia and prolonged jaundice. Motor development was delayed. Labs showed elevated liver enzymes, CK-MB, D-dimers and troponins and deficiency of growth hormone and thyroid hormones. USG scans of the brain, lungs, abdomen and heart showed only one spleen cyst. Brain MRI revealed deep interpeduncular fossa, mesencephalon and pons hypoplasia which suggested Joubert syndrome. Moreover, ectopy of posterior pituitary and suprasellar cysts were described, what together with hormones abnormalities suggested combined pituitary hormone deficiency. Supplementation of thyroid hormones and growth hormone was introduces as well as complex healthcare.

Conclusion: Described patient is an example of coexistence of two different syndromes: Joubert syndrome and combined pituitary hormone deficiency. It should remind us that making clinical diagnosis is not only simple matching present symptoms to one illness. The same symptoms may be sign of many different conditions which can co-exist in one patient. Diagnostic process should not be stopped after finding one possible justification for most of the complaints, but after disqualifying other possible explanations of theirs existence.