PROTEUS SYNDROME – rare genetic disorder in children (case raport)

Sokolova Rozeta*, Jovceski Lazo*, Lumani Njomza*

** University Clinic of Pediatric Surgery – Skopje, Macedonia*
Corresponding author: njomzalumani@gmail.com

Abstract

Proteus syndrome is a rare genetic disorder, characterised by overgrowth of various tissues. Organs and tissues affected by the disease grow out of proportion to the rest of the body. The overgrowth is usually asymmetric, which means it affects the right and left sides of the body differently. Proteus syndrome results from a mutation in the AKT1 gene. Overgrowth becomes apparent between the ages of 6 and 18 months. Progressive skeletal malformations, bullous pulmonary disease, tumors, vascular malformations and skin diseases are reported

Our patient is 2 years old female hospitalizes in our clinic with suspicion for PS, for complains for pain and clinically noted with eye swallow in the right axillary region, with restricted ROM in the right shoulder. We also noticed changes on the left forearm, and on the left small finger without inflammation. The size of the left forearm is bigger for 2 cm compared with the right forearm. The child is born with no symptoms or pathological sings. After the first year, the sings began.

We made surgical excision of the soft overgrow tissue of the thoracal wall. The result we got was fatty tissue.

We find this case interesting to show for it is very rare genetic condition with an incidence of less then 1 in 1 million people worldwide, and only a few hundred have been reported in the medical literature.

Even after surgical treatment is indicated and performed, it is necessary this children to be followed form multidisciplinary approach of different specialties pediatric, pediatric surgeon, orthopedic surgeon and genetic consultant for the expected further clinical expressions of the disease.

Keywords: Asymmetry, AKT-1 gene, Proteus