METASTATIC OSTEOBLASTIC OSTEOSARCOMA IN A PATIENT WITH HEREDITARY CANCER SYNDROME, CASE REPORT

by Zhanna Kovalova | Baiba Lace | Ksenija Soldatenkova | Zane Abola | Dzintars Ozols | University Children's Hospital | University Children's Hospital | University Children's Hospital | University Children's Hospital

Themes: Solid Tumours
Presenter Name: Zhanna Kovalova | Zhanna Kovalova
Keywords: ATM gene, hereditary predisposition, osteosarcoma

Osteosarcoma is generally considered a rare and sporadic tumor, however some cases with genetical predisposition have been confirmed. We report the case of a 15-year-old adolescent with metastatic osteoblastic osteosarcoma of diaphysis of the right femur with genetical cancer predisposition syndrome. The girl presented in hospital with symptoms of right knee swelling, dynamically progressing pain and difficulties in doing daily physical activities. The patient underwent neoadjuvant chemotherapy followed by osteoblastic osteosarcoma resection and knee joint endoprosthetic reconstruction treatment, combined with adjuvant chemotherapy courses, and local therapy of pulmonary metastases, the total amount of metastatic resected lesions in right lung was 44, on left lung 56. There was no family history of malignant diseases.

Whole exome sequencing was performed using the Illumina's sequencing-by-synthesis method, which followed by analysis of the 160 genes of Comprehensive Hereditary Cancer Panel done by private company.
Pathogenic heterozygous variant c.7630-2A>C in ATM gene was identified. GnomAD frequency of variant is low (0.001%). The variant c.7630-2A>C affects a splice acceptor site in intron 51, hence results in skipping of exon 52 and introduction a premature termination of ATM protein. It has also been described as a pathogenic founder variant in individuals of Polish descent.
The girl is in remission during last 2 years. This study presents a case of surgical arthroplastic knee joint treatment manifestation in a pediatric patient with distal femur metastatic osteoblastic osteosarcoma combined with scheduled chemotherapy courses and resection of pulmonary metastases in a patient with hereditary cancer syndrome.Osteosarcoma is generally considered a rare and sporadic tumor, however some cases with genetical predisposition have been confirmed.