

## A RARE COINCIDENCE: FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY AND BREAST CANCER

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**Aim:** Facioscapulohumeral muscular dystrophy (FSHD) is an autosomally inherited neuromuscular disorder and may be associated with increased cancer risk. **Patient:** A 69-year old female admitted to hospital with complaint of left axillary mass who had diagnosis of FSHD in her adulthood period. **Results:** Bilateral breast cancer diagnosis was made and the patient underwent bilateral mastectomy. Following the operation, adjuvant chemotherapy and radiotherapy performed and hormonal therapy started. **Conclusion:** The patients with congenital muscular dystrophy might have an increased risk of malignancy. We consider that some genetic alterations in FSHD might have contributed to the development of bilateral breast cancer in our patient.

**Key Words:** muscular dystrophy, breast cancer, PTEN.

Facioscapulohumeral muscular dystrophy (FSHD) is an autosomally inherited neuromuscular disorder which is typically characterized by the weakness of the muscles of the face, upper-arm and shoulder girdle. It has high frequency of sporadic cases. Recent studies reported that patients with myotonic dystrophy might have an increased risk of thyroid cancer and choroidal melanoma and possibly, testicular and prostate cancers [1]. A case of FSHD patient with extramedullary thoracic tumor has been reported [2]. As our knowledge we presented the first bilateral breast cancer with FSHD.

**Case report.** A 69-year old female admitted to hospital with complaint of left axillary mass. There was no other pathological finding in her physical examination. She had FSHD diagnosis in her adulthood period and she had no medication for FSHD. In her family history: her son was suffering from same disease and none of her relatives had history of breast cancer. She has been post menopausal for ten years. She did not have any breast cancer risk factors, such as hormone replacement therapy, past history of radiotherapy or smoking. Her creatine kinase levels were two times higher than normal limits (range 60–400 IU/L) and other laboratory tests were within normal limits. Mammography revealed bilateral mass in her breasts. Pathological examination of bilateral tru-cut biopsy showed invasive ductal carcinoma. Left radical mastectomy and right lumpectomy performed. Computed tomography and bone scintigraphy did not show any distant metastasis. She had bilateral invasive ductal carcinoma as T1cN2M0 grade III, ER 90% (+), PR 95% (+), c ERB2 (-) for right breast and T1c-

NOM0 grade II, ER 95% (+), PR 95% (+) and cERB2(-) for left breast. She was given adjuvant four cycles of cyclophosphamide, adriamycin (CA) sequentially radiotherapy and aromatase inhibitor.

It has been postulated that there might have been an relationship between myotonic dystrophy and cancer [3, 4]. Gadalla and colleagues reported an increased cancer risk, especially endometrium, brain, ovary and colon cancers for those who had myotonic dystrophy in a large population-based study [5].

FSHD and breast cancer pathogenesis may be related with each other. Allelic loss is one of the genetic mechanisms of breast cancer. Phosphatidylinositol-3,4,5-trisphosphate 3-phosphatase (PTEN) is a tumor suppressor gene located on chromosome 10. PTEN tumor suppressor gene mutation and loss of heterozygosity at the PTEN locus have role in breast tumorigenesis [6]. A few cases of FSHD with rearrangements between subtelomeric chromosome 4q and a subtelomeric region of 10q were reported in the literature. Gene rearrangements might lead to the loss of 10q heterozygosity and subsequently eliminating the tumor suppressor effect of PTEN on cell cycle.

In conclusion, the patients with congenital muscular dystrophy might have an increased risk of malignancy. We consider that some genetic alterations in FSHD might have contributed to the development of bilateral breast cancer in our patient. However, we need further trials to evaluate probable common genetic alterations in FSHD with malignancies. Cancer risk should be always kept in mind during follow-up of the patients with neuromuscular diseases such as FSHD.

### CONFLICT OF INTEREST

The authors declare that they have no conflict of interest.

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**Abbreviations used:** CA – cyclophosphamide – adriamycin; ER – estrogen receptor; ERB2 – avian erythroblastosis oncogene B; FSHD – facioscapulohumeral muscular dystrophy; IU/L – international unit/liter; PR – progesterone receptor; PTEN – phosphatidylinositol-3,4,5-trisphosphate 3-phosphatase; TNM – tumor – lymph nodes – metastasis.

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