

## عنوان مقاله:

Neurofibromatosis and Breast cancer

## محل انتشار:

دومین کنگره بین المللی و دهمین همایش ملی نوروزنتیک ایران (سال: 1396)

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## خلاصه مقاله:

Neurofibromatosis type1 (NF-1) or Von Recklinghausen disease is an autosomal dominant disorder with a highpenetrance and wide variability in disease expression. It has an incidence of 1:3000–4000. NF-1 is aneurocutaneous syndrome with autosomal dominant mode of inheritance and has a high propensity to developbenign and malignant nervous system tumors.The characteristic common clinical features are subcutaneousnodules (neurofibromas), café-au-lait spots, axillary, inguinal freckling, and Lisch nodules with uncommonfeatures being pseudoarthrosis, scoliosis, and parathyroid hyperplasia. Patients can develop other benign andmalignant tumors such as neurofibrosarcoma, optic gliomas, malignant schwannoma, pheochromocytoma,myelogenous leukemia, and rhabdomyosarcoma. Few case reports describe the association of breast carcinomawith NF-1. Although uncommon, case reports describing the association of NF-1 and breast cancer are availablein the literature. Since patients with NF-1 are at an increased risk of developing breast cancer, we recommendstrict adherence to careful clinical breast examination and annual screening mammographic examination startingat 40 years of age in all patients of NF-1. The age-specific excess risk of breast cancer, comparing the NF1cohort with the control cohort, was elevated 6.5-fold (95% confidence interval 2.6–13.5) in women aged 30–39years. There was a 4.4 (2.5–7.0) times higher risk among women aged 40–49

## کلمات کلیدی:

Neurofibromatosis, Breast cancer, Screening

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