MALIGNANT NEOPLASMS IN PATIENTS WITH NEUROFIBROMATOSIS TYPE 1: RETROSPECTIVE CASES REVIEWS

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AIMS

The purpose of the current study is to describe types of cancer associated with NF1 in 28 patients referred to INCA's Clinical Genetics Clinics for genetic counseling. The association between NF1 and malignant tumors has been widely described; the most common reported associations are with gliomas, malignant peripheral nerve sheath tumors, leukemia, pheochromocytoma and rhabdomyosarcoma. Concerning the association between NF1 and breast cancer, only a few cases have been reported.

PATIENTS AND METHODS

We performed a retrospective chart review of twenty-eight patients with NF1 at INCA's Clinical Genetics Clinics for genetic counseling. So far, we analyzed the age at diagnosis, family history, physical exam and evolution for cancer. .

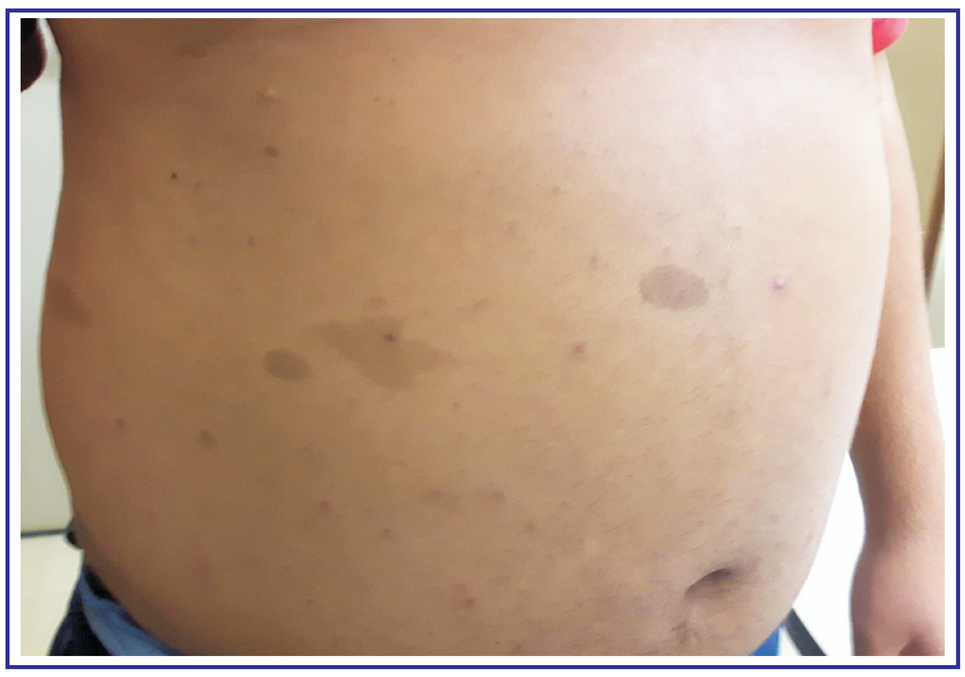
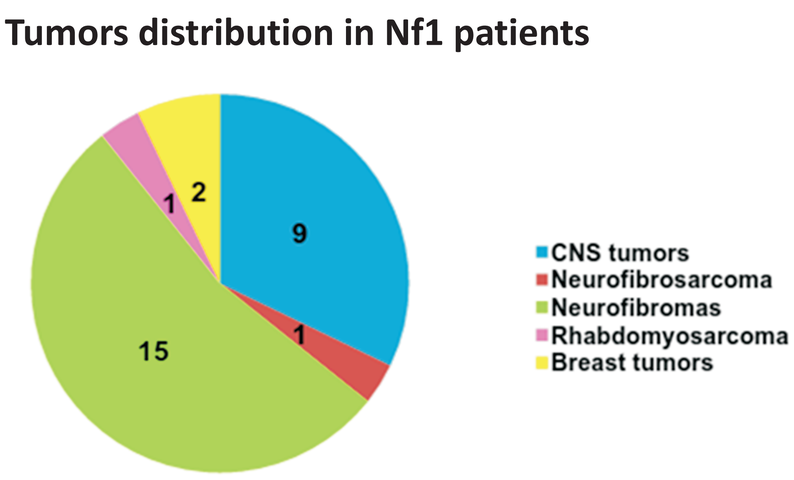


Figure 1: *café-au-lait* spots in abdomen and leg in a NF1 patient

RESULTS

The mean age of NF1 diagnosis was 64,93 months (SD: 63,18 months, minimum 1 month and maximum 192 months of age). Eleven patients were diagnosed with central nervous system tumor; 5 with optic gliomas, 2 astrocytoma, 2 posterior fossa tumor, 1 medial fossa tumor and 1 mesencephalon, thalamus and optic pathways. We identified a total of four different tumors types: malignant astrocytoma (2), neurofibrosarcoma (1), rhabdomyosarcoma (1) and breast cancer (2). Regarding to family history, 2 patients had first degree relatives with breast cancer.



CONCLUSION

The types of cancer found in our sample were similar to those found in the medical literature. Even if the association between breast cancer and NF1 is rarely reported, the few studies found in the literature suggest that women with NF1 are at a higher risk of developing breast cancer when compared to the general population. Interestingly, *NF1* gene and *BRCA1* gene are both located in the peri-centromeric region of the long arm of chromosome 17 and about 28% of sporadic breast cancers are missing at least one copy of the *NF1* gene, either due to deletion or mutation. The findings of the above-mentioned reports and other published data justify the requirement of specific screening programs for breast cancer in NF1 patients. Moreover, cancer management in this population is not well defined; especially when some available data suggests that the risks of fibrosarcomas are increased by radiation when a conservative approach is chosen for this population. Counseling of patients and their families should provide a realistic overview of possible clinical complications, while emphasizing that most individuals with NF1 have healthy and productive lives.

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Meeting: XXIX CONGRESSO BRASILEIRO GENÉTICA MÉDICA 2017

Financial support: CNPq, CAPES

Projeto Gráfico: Serviço de Edição e Informação Técnico-Científica / INCA

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