

Recent advances in our knowledge of hereditary breast cancer

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Hereditary breast cancer: an update on genotype and phenotype.
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<http://www.geocities.com/m.lacroix/livre4.htm>

A significant minority (5-10%) of breast carcinomas is considered of hereditary origin. Part of them is associated to mutations in the BRCA1 and BRCA2 susceptibility genes, which have been extensively studied. For their part, tumours in the non-BRCA1/BRCA2 breast cancer (BRCAx) families are increasingly believed to originate from multiple genetic events, as no "BRCA3" gene has been identified. The existence of specific "portraits" among hereditary breast carcinomas (HBC) has been questioned. Phenotype studies have shown that most BRCA1 tumours are part of a "basal epithelial-like" group of lesions. In contrast, BRCA2 and BRCAx HBC are more heterogenous, as also observed with sporadic carcinomas. Genetic analysis has also been applied to HBC, notably with the objective of resolving the heterogeneity of BRCAx lesions. This chapter aims to summarise data on BRCA1, BRCA2, and BRCAx HBC, and to provide hypotheses on the origin of BRCA1 lesions, their high-frequency of P53 mutations, and their paradoxical relations to oestrogen-sensitivity.

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