

DISEASE:
Hereditary breast cancer

NAME:	Hereditary breast cancer
DESCRIPTION:	A rare genetic, malignant breast tumor characterized by early onset breast cancer in association with a germline mutation. Tumors arising in carriers of BRCA1 and BRCA2 mutations differ morphologically and genetically from each other, as well as from sporadic breast cancers. Most BRCA1-associated tumors are invasive ductal adenocarcinomas of no special type, typically of higher grade than sporadic tumors, and more often negative for hormone receptors. In addition, more cases with features of typical or atypical medullary carcinoma are seen in these patients. Likewise, BRCA2-associated tumors tend to be of higher grade than sporadic ones, although their phenotype is similar. They show a low frequency of HER-2 expression.
ORPHACODE:	227535
SYNOMYS:	Familial breast cancer Familial breast carcinoma Hereditary breast carcinoma

XREF(S):	Orphanet ICD-10 ICD-10 ICD-10 ICD-10 ICD-10 ICD-10 OMIM OMIM OMIM OMIM ICD-10
ANALYTE(S):	CDH1 BRCA1 BRCA2 PALB2 XRCC2 KLLN RAD54L PPM1D SLC22A18
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