



CDH1 WOMEN



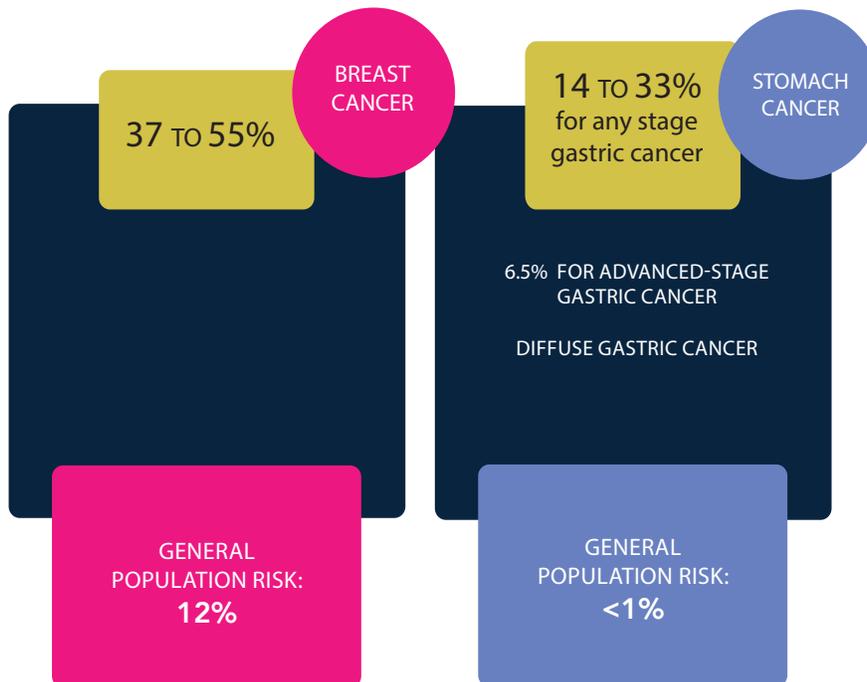
The medical management guidelines from the National Comprehensive Cancer Network (NCCN) for patients with a CDH1 pathogenic / likely pathogenic variant are listed in this document.

This overview is for informational purposes and does not constitute a personalised recommendation. **Recommended options may vary based on your personal and family history. Access to some options may also vary from one medical center to another.** The specific references should be consulted for more details before developing a treatment plan.

In addition, the information available on hereditary cancer susceptibility genes is constantly evolving and **it is recommended to check this information annually as the management guidelines may change in the future.**

LIFETIME ASSOCIATED RISKS

(UP TO AGE 75)



NOTE

Cleft lip with or without cleft palate has been associated with CDH1 pathogenic and likely pathogenic variants.

References:

Daly M et coll. NCCN Clinical Practice Guidelines in Oncology: Genetic/Familial High-Risk Assessment: Breast, Ovarian and Pancreatic. Version 3.2025-March 6, 2025. <http://www.nccn.org>

Barbosa-Matos R, Córdova L, Schrader K, et al. Diffuse Gastric and Lobular Breast Cancer Syndrome. 2002 Nov 4 [Updated 2024 Oct 10]. In: Adam MP, Feldman J, Mirzaa GM, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2025. <https://www.ncbi.nlm.nih.gov/books/NBK1139/>

Gupta S et coll. NCCN Clinical Practice Guidelines in Oncology: Genetic/Familial High-Risk Assessment: Colorectal, Endometrial, and Gastric. Version 4.2024. April 2, 2025. <http://www.nccn.org>



CDH1 WOMEN



BREAST CANCER

STOMACH CANCER

SCREENING

STARTING AT AGE 18

IF ELECT TO NOT UNDERGO PROPHYLACTIC TOTAL GASTRECTOMY

- UPPER ENDOSCOPY EVERY 6 TO 12 MONTHS WITH MULTIPLE RANDOM BIOPSIES

IMPORTANT : THE EFFECTIVENESS OF THIS SCREENING METHOD HAS NOT BEEN RECOGNIZED FOR THE DETECTION OF EARLY GASTRIC CANCER

STARTING AT AGE 75

- MANAGEMENT SHOULD BE DETERMINED ON AN INDIVIDUAL BASIS

18

30

75

STARTING AT AGE 30

- MAMMOGRAM WITH EVERY 12 MONTHS
- CONSIDER BREAST MRI WITH CONTRAST EVERY 12 MONTHS

STOMACH CANCER

RISK REDUCTION

SURGERY

BETWEEN AGES 18 AND 40 :
OR EARLIER ACCORDING TO FAMILY HISTORY OF GASTRIC CANCER PRIOR TO AGE 25

- PROPHYLACTIC TOTAL GASTRECTOMY (REMOVAL OF THE STOMACH BEFORE CANCER DEVELOPS) IS RECOMMENDED IF ANY OF THE FOLLOWING ARE PRESENT:
 - ESTABLISHED STAGE PT1B OR HIGHER SRCC (SIGNET-RING CELL CARCINOMA)
 - PERSISTENT SIGNS AND SYMPTOMS ASSOCIATED WITH SRCC, SUCH AS : WEIGHT LOSS, EARLY SATIETY, ANEMIA AND ABDOMINAL PAIN
- A BASELINE ENDOSCOPY IS INDICATED PRIOR TO PROPHYLACTIC TOTAL GASTRECTOMY.
- A D2 LYMPH NODE DISSECTION IS NOT NECESSARY FOR PROPHYLACTIC TOTAL GASTRECTOMY.

IMPORTANT : THE HISTOLOGY ANALYSIS OF THE BIOPSIES SHOULD BE THOROUGH TO RULE OUT THE PRESENCE OF MACROSCOPIC TUMOR FOCI OR OTHER FACTORS THAT MAY MODIFIED THE SURGERY.

BREAST CANCER

RISK REDUCTION

SURGERY

PROPHYLACTIC BILATERAL MASTECTOMY (REMOVAL OF THE BREASTS BEFORE CANCER DEVELOPS), WITH OR WITHOUT RECONSTRUCTION, HAS BEEN SHOWN TO REDUCE THE RISK OF DEVELOPING BREAST CANCER BY UP TO 90%.