

“Right to Innovation: Understanding Personalised Prevention”

Unmet Needs in Oncology – Personalised Prevention
Hereditary Cancer Syndromes

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[Evita Cancro – Evitar o Cancro hereditário](#)

ePAG [ERN GENTURIS](#)



Overview

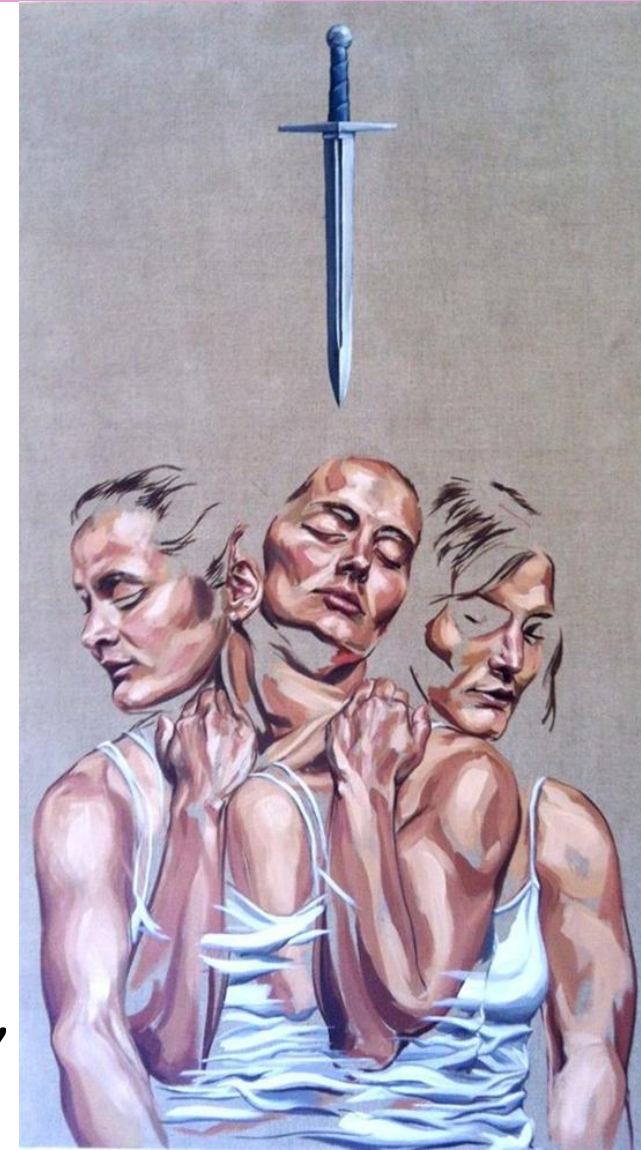


- Unmet Needs
- Solution
- How WE can help
- How YOU can help
- Take home message

Since 2011, EVITA was globally the first patient association supporting families affected by all hereditary cancer syndromes

FACTS AND UNMET NEEDS

- **Hereditary Cancer (HC)** is a consequence of a **genetic predisposition** due to an **inherited pathogenic germline variant** (commonly known as “mutation”), rare “de novo”.
- The genetic variant carrier (GVC) has an elevated risk to develop **early-onset cancer in a group of organs**: BRCA2 carriers have an increased risk for breast, ovarian, prostate and pancreatic cancer.
- Each cancer is potentially involved in a **panel of genes**. In case of **breast cancer**: BRCA1/2, PALB2, PTEN, STK11, TP53, CHEK2, ATM, BARD1, BRIP1 or RAD51, e.g., besides others.



FACTS and UNMET NEEDS

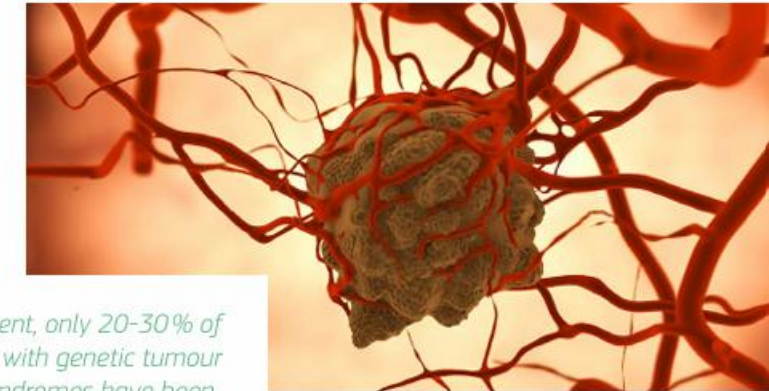
- HC represents **10% ???** of all cancers.
- Only **20%** of GVC are identified – **80%** are not aware of their genetic predisposition!
- **Over 50%** of HC patients do not meet testing criteria by current guidelines or do not undergo genetic testing.

[Few People with Cancer Undergo Germline Testing – NCI](#)

ERN on genetic tumour risk syndromes (ERN GENTURIS)

Genetic tumour risk syndromes are disorders in which inherited genetic mutations strongly predispose individuals to the development of tumours. The lifetime risk of cancer can be as high as 100%. While there is considerable diversity in the organ systems that may be affected, individuals affected by these conditions share similar challenges: delay in diagnosis, lack of prevention for patients and healthy relatives, and therapeutic mismanagement. At present, only 20-30% of people with genetic tumour risk syndromes have been diagnosed.

ERN GENTURIS is working to improve identification of these syndromes, minimise variation in clinical outcomes, design and implement guidelines, develop registries and biobanks, support research, and empower patients. The network will educate the public and healthcare professionals, and foster the sharing of best practice across Europe. Access to multidisciplinary care will be improved, with new models and standards for sharing and discussing complex cases. The network is enhancing the quality and interpretation of genetic testing,



At present, only 20-30% of people with genetic tumour risk syndromes have been diagnosed.

and increasing patient participation in clinical research programmes.

ERN GENTURIS will cooperate with other ERNs to improve the care of patients with genetic tumour risk syndromes who develop conditions that fall within the expertise of another network.

NETWORK COORDINATOR

Prof. Nicoline Hoogerbrugge
Radboud University Medical
Center Nijmegen, The Netherlands

FACTS and UNMET NEEDS



Statistics at a glance, 2022

New cancer cases in **Portugal** in 2022: 70.000 – **7.000/year, 19/day**

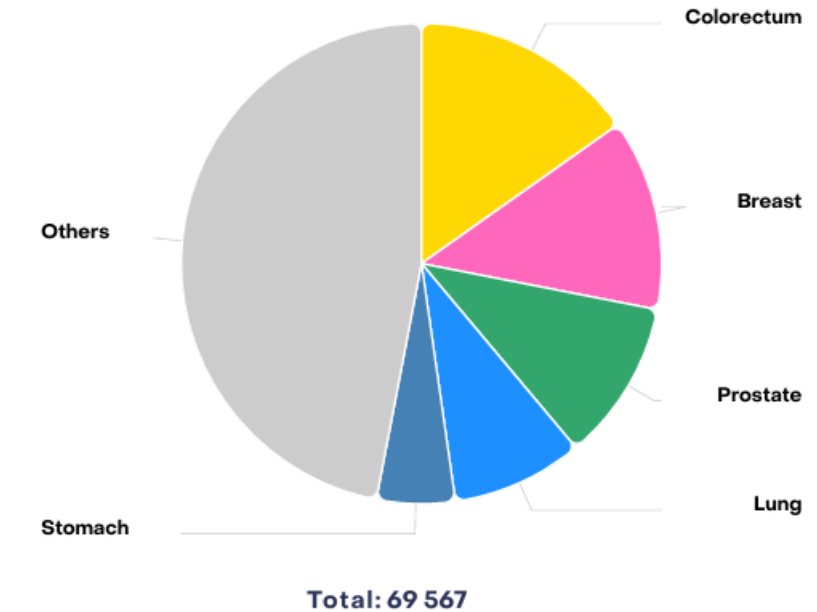
Europe: **450.000/year, 1233/day**

The 3 most prevalent cancers (38.9%) are potentially involved within the 2 most frequent HCS: HBOPC and Lynch Syndrome.

Lung cancer (47,7%) recently also linked to HCS

[Inherited lung cancer: a review - PMC](#)

Both sexes



Rank	Cancer site	Number of cases	Percent
1st	Colorectum	10 575	15.2%
2nd	Breast	8 954	12.9%
3rd	Prostate	7 529	10.8%
4th	Lung	6 155	8.8%
5th	Stomach	3 668	5.3%
-	Others	32 686	47.0%

FACTS AND UNMET NEEDS



- If GVC and/or HCP are not aware of the genetic predisposition, **first symptoms may be disregarded** – delay of diagnosis.
- Due to early-onset of HC, during the **peak of fertility and productivity**, and quite often late-stage diagnosis, HC represents a **high socioeconomic burden** (**numbers are about to be published by EU Project [Home - Preventable](#)**).
- **Low genetic literacy** in civil and even medical society blocks access to gc+gt
- **Complexity hinders** identification of patterns and symptoms.
- Dispersed information.

FACTS AND UNMET NEEDS



- Family matter: Impact on **mental health** within the family remains underserved.
- Unacceptable **waiting times** for genetic counseling – genetic testing (**precision diagnosis**) – **precision screening** - **precision prevention** by risk-reducing surgeries and PGD.
- **Inequity** in treatment access – **precision treatment**.
- **Lack of strategy and low budget** on prevention and early detection.
- Recently updated **European Code Against Cancer** continues to overlook the highest risk factor: **genetic predisposition!**

Unmet Needs in Hereditary Cancer Syndromes

- Portuguese **Founder Mutation**

BRCA2 c.156_157insAlu (*learn more)

30% of all BRCA related cancers

60% of all BRCA2 related cancers

Populational screening for females > 25 y

- **No data**

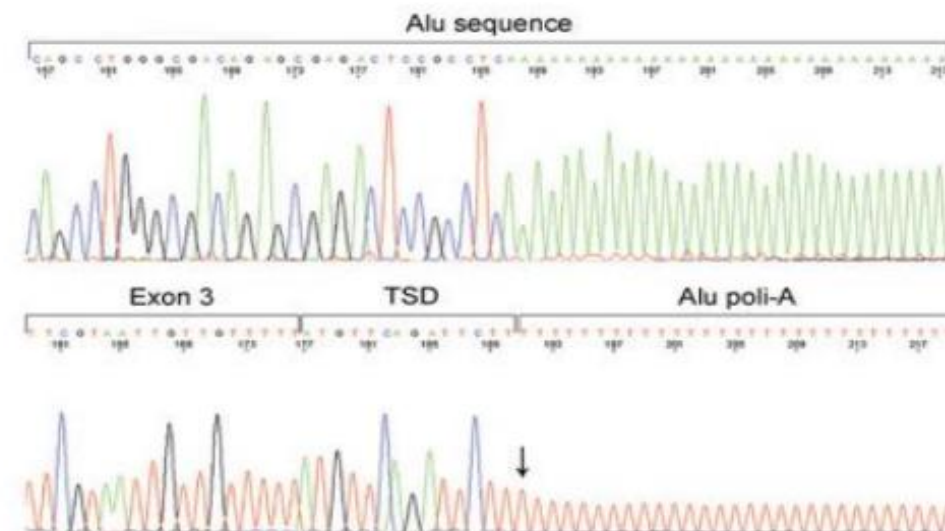
- **Lack of PPIE** in HTA and research

*[Registration \(gotowebinar.com\)](http://gotowebinar.com)

Founder pathogenic variant of Portuguese origin

BRCA2 c.156_157insAlu is a founder pathogenic rearrangement of Portuguese origin identified in 8% of index cases and representing 60% of *BRCA2* alterations in individuals of Portuguese ancestry. Its presence causes an in-frame deletion of exon 3 and it is deleterious. Its identification might be challenging in case it is not being proactively analyzed.

We would like to raise awareness about this alteration and recommend their analysis in families with clinical suspicion of hereditary breast and ovarian cancer and a Portuguese ancestry.



For more technical details, please follow this link: [BRCA2 c.156_157insAlu](#)

National and European Policies



"The NCP's Hereditary Cancer Working Group, while focused on early detection, is challenged to improve its approach to hereditary cancer prevention, with an emphasis on identifying, monitoring, and managing risk in families with a genetic predisposition."



www.genturis.eu



EARLYSCAN Cluster:
SHIELD, DISARM, PREDI-LYNCH

Solution: EVITA Platform, 3 user profiles

Citizen to Citizen Initiative



evita
PLATFORM

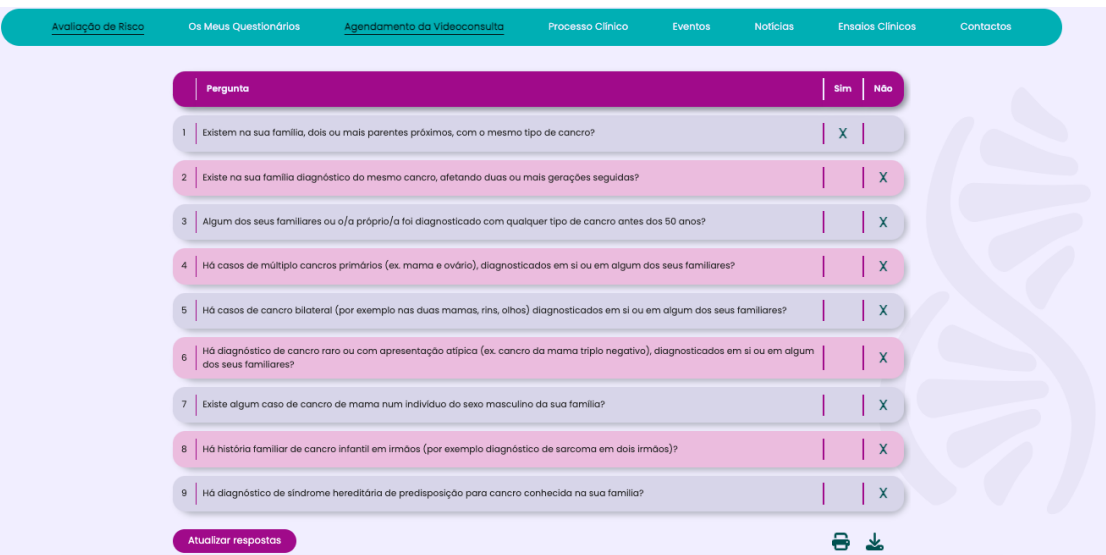
A EVITA Platform é uma ferramenta digital que visa apoiar os cidadãos preocupados com o seu risco para cancro, os portadores de alterações genéticas com risco para cancro hereditário e todos os doentes oncológicos

Utilizador EVITA	Profissional de Saúde	Investigador
		
Bem-Vindo	Bem-Vindo	Bem-Vindo
Entrar	Entrar	Entrar
Inicie sessão ou registe-se	Autentique-se	Investigação na EVITA Platform
Registe-se aqui	Apenas médicos oncologistas, geneticistas e psicólogos terão acesso a esta área	Registe-se aqui

We provide a **Familial Cancer Risk Awareness Tool** that generates an immediate result:
Short questionnaire based on national recommendations PROGO



Cancro Hereditario



The screenshot shows a web interface for a questionnaire. At the top, there is a navigation bar with the following items: Avaliação de Risco, Os Meus Questionários, Agendamento da Videoconsulta, Processo Clínico, Eventos, Notícias, Ensaio Clínico, and Contactos. The main content area is titled 'Pergunta' and contains a list of 9 questions. Each question has a 'Sim' (Yes) and 'Não' (No) column with a radio button. The 'X' mark is visible in the 'Sim' column for questions 1, 2, 3, 4, 5, 6, 7, 8, and 9. At the bottom of the questionnaire, there is a button labeled 'Atualizar respostas' and two icons: a printer and a download arrow.

Pergunta	Sim	Não
1 Existem na sua família, dois ou mais parentes próximos, com o mesmo tipo de cancro?	X	
2 Existe na sua família diagnóstico do mesmo cancro, afetando duas ou mais gerações seguidas?		X
3 Algum dos seus familiares ou o/a próprio/a foi diagnosticado com qualquer tipo de cancro antes dos 50 anos?		X
4 Há casos de múltiplo cânceros primários (ex. mama e ovário), diagnosticados em si ou em algum dos seus familiares?		X
5 Há casos de cancro bilateral (por exemplo nas duas mamas, rins, olhos) diagnosticados em si ou em algum dos seus familiares?		X
6 Há diagnóstico de cancro raro ou com apresentação atípica (ex. cancro da mama triplo negativo), diagnosticados em si ou em algum dos seus familiares?		X
7 Existe algum caso de cancro de mama num indivíduo do sexo masculino da sua família?		X
8 Há história familiar de cancro infantil em irmãos (por exemplo diagnóstico de sarcoma em dois irmãos)?		X
9 Há diagnóstico de síndrome hereditária de predisposição para cancro conhecida na sua família?		X



The screenshot shows the result page of the questionnaire. At the top, there is a navigation bar with the following items: Avaliação de Risco, Os Meus Questionários, Área Clínica, Agendar Videoconsulta, Eventos, Notícias, Ensaio Clínico, and Contactos. The main content area is titled 'RESULTADO' and contains the following text: 'Os resultados do teste que acabou de realizar indicam que o risco para cancro hereditário na sua família aparenta ser inconclusivo. Recomenda-se uma consulta com um médico-geneticista, que o irá ajudar a fazer uma melhor avaliação e fornecer-lhe-á todas as informações adicionais que necessita.' Below this, there is a section titled 'IMPORTANTE' with the following text: 'O teste deve ser realizado novamente sempre que obtenha mais informações acerca de casos de cancro na sua família (designadamente, casos de diagnóstico de cancro nos seus familiares que desconhecia no momento do teste, casos de cancro na sua família que foram diagnosticados após o teste).' At the bottom of the result page, there is a button labeled 'Ver Respostas'.

We do NOT diagnose anything, but we can **help to understand** citizens and hcp´s
WHO could benefit from genetic counseling

GDPR compliant Digital Platform with high standard technology for EVERYONE

“User EVITA” Profile – Concept: Users are in charge of their health and their data!

For any citizen concerned about their cancer risk, carriers of genetic variants, and all cancer patients:

- **Familial Cancer Risk Awareness Questionnaire**
- **Health literacy**, lay summaries, newsletters, webinars
- **Clinical trials** recruiting in Europe



- **Clinical area:** a) Secure space for uploading the entire clinical process in a centralized manner to overcome the barrier of system **interoperability** and to **prevent waste** (duplication of exams and analyses...), and
b) a **clinical diary** with the possibility of granting access to the authorized oncologist = the doctor can better prepare for the next consultation, optimizing the limited time (**telemonitoring**).
- **Digital agenda** to book appointments.
- **Telemedicine:** Genetic counseling and psycho-oncological consultation.
- **Digital Tool** “My Questionnaires” designed for **active participation in research & HTA**
- **APP**

EVITA Platform respects **FAIR principles** and aims to integrate the **EHDS**

User profiles "Healthcare Professional" and "Researcher"

Oncologist:

- With prior authorization, will have access to the information selected in the clinical area by the user
- Communication channel (clinical diary)

Geneticist:

- Reserved area for Video Consultation for Genetic Counseling, shortening the waiting time

Psychologist:

- Reserved area for Video Consultation

Researcher:

- Research questions with target audience selection
- Report output with structured data
- Access to the reservoir of previous research
- PROMs, PREMs, RWD, RWE



The screenshot displays the Evita platform's user interface. At the top, a navigation bar includes links for 'Avaliação de Risco', 'Os Meus Questionários', 'Área Clínica', 'Agendar Videoconsulta', 'Eventos', 'Notícias', 'Ensaio Clínicos', and 'Contactos'. Below this, there are two main panels. The left panel, titled 'Suspeita que há uma predisposição para cancro na sua família?', contains a questionnaire prompt, a 'Continuar' button, and two additional links: 'Clique aqui para completar o seu perfil EVITA!' and 'Clique aqui para configurar o seu processo clínico'. The right panel, titled 'Profissionais de saúde', is for 'Permissões de Acesso aos Dados Demográficos e Clínicos' and features an 'Associar Profissional de Saúde:' section with a text input field for a professional's code and an 'Adicionar' button. A note at the bottom of this panel states: 'Ao associar o profissional de saúde, este poderá aceder aos seus dados do processo clínico de acordo com cada perfil médico. Pode restringir o acesso através da alteração das permissões abaixo.'



Encryption of clinical documents that the user uploads to the platform:

- Physically stored on the Azure server.
- Encrypted twice using the AES algorithm
- 1st encryption is done with a key unique to all users and then encrypted again with a unique key/code per user.
- This unique code for each user is used to decrypt the passwords and documents (this unique code is generated during registration).
- The decrypted documents can be accessed by the user and by any healthcare professionals the user authorizes.

Access and storage of personal data (demographic data):

- The database is stored on an isolated server (protected by credentials), not exposed to the internet, and only available to application services on the same network.
- Remote access to virtual machines is restricted to specified IP addresses.
- Accessed only by the user themselves;
- Accessed by doctors authorized by the user.
- The platform users' passwords are encrypted.
- All HTTP traffic (activities on the EVITA Platform: such as entering the "clinical area") is securely conducted using SSL (Security Socket Layer) certificates.

Note of Thanks

Parceiros Platinum



Parceiros Silver



Parceiros Gold



Donativos



Prémios



Legal Advisor: Cláudia Monge

EVITA DREAM TEAM

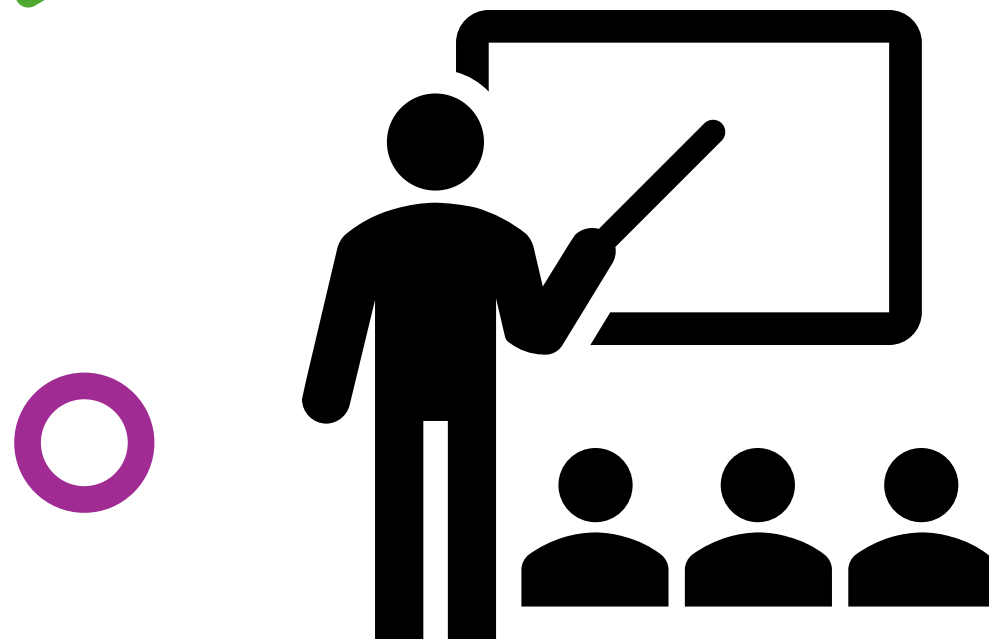
Catarina Ferreira (Scientific Advisor)
Tatiana Vassilevskaia (Project Manager EP)
Laura Carreto (Int. Project Manager)
Mariana Coutinho (Executive Director)
Tamara Milagre (President)

How YOU can help

To reinforce the commitment to collaboration:

GROW THE FLOW

- Dissemination & recommendation (D & R) of EVITA Platform at national and international patient, medical and scientific events.
- D & R during medical consultations in General Practise, Oncology, Medical Genetics, and other interconnected specialties.
- D & R in waiting rooms (brochures and posters with QR codes)
- Spread the word in your communities and with decision makers and other relevant stakeholders!
- We aim the formal recognition by Health Entities like the Ministry of Health and Medicines Agency within a strategic partnership.
- We count on your endorsement!



Take Home Message

Prevention of waste and disease are fundamental pillars for the sustainability of the system

HCS are actionable

Identify more and better genetic variant carriers

Use available digital tools like EVITA Platform to facilitate

Organise ideal pathways within the system (PREVENTABLE)

Healthier and happier families

We can save more lives together!



[EVITA x Da Weasel \(youtube.com\)](https://www.youtube.com/watch?v=EVITAxDaWeasel)



DISCOVER CANCER
BEFORE IT'S CANCER

Cancer is the biggest opponent you may ever face. When you confront it for the first time, it seems invincible. It's a tough battle, with advances and setbacks. And in the end, even if you win, you won't be the same person again. But did you know that cancer can be a hereditary disease? And that, currently, about 10% of cancer cases are of hereditary origin? If you have a family history of cancer, visit evitaplatform.org and assess your cancer risk based on genetic predisposition. Take preventive action. If you discover it before it exists, you may not have to face it.

FIND OUT MORE ON
EVITACANCRO.ORG



a PeRsOnalized Prevention roadmap
for the future HEalthcare

THANK YOU FOR YOUR ATTENTION!

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[EVITA Platform](#)

For further informations regarding EVITA Platform,
please reach out to

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