

# ERN GENTURIS PLAIN LANGUAGE SUMMARY OF GUIDELINE ON COUNSELLING ON REPRODUCTIVE OPTIONS FOR INDIVIDUALS WITH A CANCER PREDISPOSITION SYNDROME (INCLUDING GENTURIS)

**Guideline authors:** Said C. Farschtschi, Candy Kumps, Tamara Hussong Milagre, ERN GENTURIS counselling on reproductive options guideline group\*, Sandra Janssens, Sarah Pugh, Laura Kirstine Sønderberg Roos.

\*Core working group members in alphabetical order:

Author	Speciality/Role	Affiliation
Said C. Farschtschi, MD	Physician, registrar for neurology and genetic counselling	University Medical Center Hamburg-Eppendorf, Hamburg, Germany; Member of ERN GENTURIS
Sandra Janssens, MD, PhD	Clinical geneticist	University Hospital Ghent, Ghent, Belgium; Member of ERN GENTURIS
Candy Kumps, MD, PhD	Clinical geneticist	University Hospital Ghent, Ghent, Belgium; Member of ERN GENTURIS
Tamara Hussong Milagre	Community representative	EVITA Association – Hereditary Cancer (Associação EVITA – Cancro Hereditário, Portugal; ePAG of ERN GENTURIS
Sarah Pugh	Consultant Genetic counsellor	Manchester Centre for Genomic Medicine, Manchester University Foundation NHS trust, Manchester, United Kingdom
Laura Kirstine Sønderberg Roos, MD, PhD	Clinical geneticist (chair)	Rigshospitalet, Copenhagen, Denmark; Member of ERN GENTURIS

\*Other members in alphabetical order:

Author	Speciality/Role	Affiliation
Anna Sophie Berghoff, MD, PhD	Oncologist	Medical University of Vienna, Vienna, Austria; Member of ERN GENTURIS
Estela Carrasco López, MSc	Genetic counsellor	Vall d'Hebron University Hospital, Barcelona, Spain; Member of ERN GENTURIS
Claudia Cesaretti, MD	Clinical geneticist	Fondazione IRCCS Ca' Granda, Ospedale Maggiore Policlinico, Milan, Italy; Member of ERN GENTURIS
Ellen Denayer, MD, PhD	Clinical geneticist	University Hospitals Leuven, University of Leuven, Leuven, Belgium; Member of ERN GENTURIS
Francesca Fianchi, MD	Internist	Fondazione Policlinico Universitario A. Gemelli IRCCS, Rome, Italy; Member of ERN GENTURIS
Marianne Geilswijk, MD	Clinical geneticist	Aarhus University Hospital, Aarhus, Denmark; Member of ERN GENTURIS
Mariëtte van Kouwen, MD, PhD	Gastroenterologist	Radboud university medical center, Nijmegen, the Netherlands; Member of ERN GENTURIS
Periklis Makrythanasis, MD, PhD	Medical geneticist	'Aghia Sophia' Children's Hospital, National and Kapodistrian University of Athens, Athens, Greece; Member of ERN GENTURIS University of Geneva, Geneva, Switzerland Biomedical Research Foundation of the Academy of Athens, Athens, Greece
Renata d' Oliveira	Clinical geneticist	Unidade Local de Saúde (ULS) São João, Porto, Portugal; Member of ERN GENTURIS
Claas Röhl	Community representative	NF Kinder/ NF Patients United, Vienna, Austria; ePAG of ERN GENTURIS
Diana Salinas-Chaparro, MSc	Genetic counsellor	Hospital Sant Joan de Deu, Barcelona, Spain; Member of ERN GENTURIS
Ileen Slegers, MSc	Genetic counsellor, midwife specialist	UZ Brussel, Brussel, Belgium; Member of ERN GENTURIS
Irene Spinelli, MD	Gastroenterologist	Fondazione Policlinico Universitario A. Gemelli IRCCS, Rome, Italy; Member of ERN GENTURIS
Manon Suerink, MD, PhD	Clinical geneticist	Leiden University Medical Center, Leiden, the Netherlands; Member of ERN GENTURIS
Salvo Testa	Community representative	Fondazione Mutagens (hereditary syndromes carriers), Milano, Italy
Ariane Van Tongerloo	Psychologist	University Hospital Ghent, Ghent, Belgium; Member of ERN GENTURIS
Eva Trevisson, MD, PhD	Clinical geneticist	University of Padua, University Hospital of Padua, Padua, Italy; Member of ERN GENTURIS

**Disclaimer:** The content of this plain language summary is based on the "ERN GENTURIS GUIDELINE ON COUNSELLING ON REPRODUCTIVE OPTIONS FOR INDIVIDUALS WITH A CANCER PREDISPOSITION SYNDROME (INCLUDING GENTURIS)\_ final version\_august2025".

## INTRODUCTION

Cancer predisposition syndromes are genetic conditions, that can cause multiple concerns for affected family members: Not only is there an increased risk of developing cancer compared to the general population - many individuals with these syndromes also have a high risk of having a child with the same condition, which makes the process of family planning more complex. Moreover, other non-tumorous manifestations may affect clinical care as well as counselling.

The family therefore relies on health care professionals to inform and guide them through the many options that are available. However, not all healthcare professionals have the specialised knowledge needed to offer this counselling.

## GUIDELINE AIMS

The aim of this guideline is to assist healthcare professionals in providing both relevant and timely counselling for individuals with a cancer predisposition syndrome.

## SUMMARY

The guideline group consist of experts with experience in multiple areas surrounding the care of individuals with cancer predisposition syndromes as well as individuals with a cancer predisposition syndrome and their representatives. The following recommendations regarding reproductive counselling are made based on scientific literature and expert consensus obtained via a modified Delphi process.

It is recommended that all individuals with a cancer predisposition syndrome, and those family members for whom it is relevant, are offered counselling about their options with respect to family planning – it is up to them to accept or decline the offer. It is also recommended that there are several opportunities for counselling throughout a person's life, as perspectives and relevance regarding family planning can change over life.

## KEY RECOMMENDATIONS / GUIDELINE SUMMARY

Reproductive decision making - content and framework of reproductive counselling	Recommen- dation	Strength
Reproductive counselling should be offered to all individuals with a cancer predisposition syndrome* and relevant family members.	1,2, 11	Strong (1,2), moderate (11)
Couples should have access to a multidisciplinary team of healthcare experts.	6, 13	Strong

Timing of reproductive counselling provision		
Reproductive counselling should be offered longitudinally with multiple opportunities for counselling throughout life, ideally starting before family planning	7, 8	Strong
Children at risk should be offered counselling once they reach adulthood, or earlier if appropriate	10	Moderate
Presentation of reproductive option		
Reproductive counselling should provide follow-up opportunities, and access to psychological support.	12, 13	Moderate (12), strong (13)
Range of assisted reproductive technologies		
Fertility preservation options should be included in reproductive counselling.	15, 16	Strong (15), moderate (16)

\* Counselling is especially relevant in the reproductive age but can be relevant in other age group as well, such as adolescence and older individuals informing their relatives.

## PSYCHOLOGICAL NEEDS

It is important to consider the impact of a cancer predisposition syndrome on both mental and social wellbeing. A delayed diagnosis, uncertainty about future health problems and/or fear of developing cancer may cause anxiety or depression. Living with a long-term health condition may also have social challenges. There may be financial concerns such as cost of insurance and impact on employment. There may also be feelings of guilt and concerns when making plans for starting a family.

Addressing the psychological needs of patients and families with cancer predisposition syndromes should form a key element of their care and should also be part of the genetic counselling process. Healthcare professionals should ask about wellbeing at each clinical contact and be aware of signs of anxiety and depression. Patients should be referred for professional support if needed. Peer-to-peer support through patient support groups can also play a key role in wellbeing.