

Alma Mater Studiorum Università di Bologna
Archivio istituzionale della ricerca

Decision making and experiences of young adults undergoing presymptomatic genetic testing for familial cancer: A longitudinal grounded theory study

This is the final peer-reviewed author's accepted manuscript (postprint) of the following publication:

Published Version:

Godino, L., Jackson, L., Turchetti, D., Hennessy, C., Skirton, H. (2018). Decision making and experiences of young adults undergoing presymptomatic genetic testing for familial cancer: A longitudinal grounded theory study. EUROPEAN JOURNAL OF HUMAN GENETICS, 26(1), 44-53 [10.1038/s41431-017-0030-1].

Availability:

This version is available at: <https://hdl.handle.net/11585/625522> since: 2018-02-26

Published:

DOI: <http://doi.org/10.1038/s41431-017-0030-1>

Terms of use:

Some rights reserved. The terms and conditions for the reuse of this version of the manuscript are specified in the publishing policy. For all terms of use and more information see the publisher's website.

This item was downloaded from IRIS Università di Bologna (<https://cris.unibo.it/>).
When citing, please refer to the published version.

(Article begins on next page)

This is the peer reviewed accepted manuscript of:

Godino L, Jackson L, Turchetti D, Hennessy C, Skirton H.

Decision making and experiences of young adults undergoing presymptomatic genetic testing for familial cancer: a longitudinal grounded theory study.

Eur J Hum Genet. 2018 Jan;26(1):44-53.

The final published version is available online at: <https://doi.org/10.1038/s41431-017-0030-1>

Rights / License:

The terms and conditions for the reuse of this version of the manuscript are specified in the publishing policy. For all terms of use and more information see the publisher's website.

This item was downloaded from IRIS Università di Bologna (<https://cris.unibo.it/>)

When citing, please refer to the published version.

1 **TITLE PAGE**

2 **Title:**

3 Decision-making and experiences of young adults undergoing presymptomatic genetic
4 testing for familial cancer: a longitudinal grounded theory study

5 **Running title:**

6 Presymptomatic genetic testing in young adults

7 **Authors:**

8 Lea Godino^{1,2}, Leigh Jackson², Daniela Turchetti¹, Catherine Hennessy³, Heather
9 Skirton²

10 1 Center for Studies on Hereditary Cancer, Department of Medical and Surgical
11 Sciences, University of Bologna, and Unit of Medical Genetics, S.Orsola-Malpighi
12 Hospital, Bologna, Italy

13 2 School of Nursing and Midwifery, Faculty of Health and Human Sciences, Plymouth
14 University, Plymouth, UK

15 3 Visiting Professor, Faculty of Health and Social Sciences, School of Health and Social
16 Care, Bournemouth University, Bournemouth, UK

17

18 **Correspondence to:**

19 Lea Godino,
20 U.O. Genetica Medica
21 Dipartimento di Scienze Mediche e Chirurgiche
22 Policlinico Sant'Orsola-Malpighi
23 Padiglione 11
24 via Massarenti, 9
25 40138 Bologna - Italy
26 Phone +39 051 2088425
27 Fax +39 051 2088416
28 e-mail: lea.godino@studio.unibo.it

29

30 **Disclaimers:**

31 The authors have no conflicts of interest

32

33 **Abstract**

34 Enabling informed choice is an essential component of care when offering young adults
35 presymptomatic testing for a genetic condition. A systematic review on this topic
36 revealed that many young adults grew up with little information regarding their genetic
37 risk and that parents had applied pressure to them during the testing decision-making
38 process. However, none of the studies retrieved were conducted in South-European
39 countries. To address this gap, we undertook a qualitative study based on grounded
40 theory to explore the psychosocial implications of presymptomatic testing for hereditary
41 cancer in Italian young adults aged 18-30 years. Interviews were conducted on three
42 occasions: one month before counselling, and two weeks and six months after results.
43 Data were coded and grouped under themes. A total of 42 interviews were conducted.
44 Four themes emerged: knowledge, genetic counselling process, decision-making and
45 dealing with test results. Although participants grew up with little or no information
46 about their genetic risk, none expressed regret at having the test at a young age. Pre-test
47 counselling was appreciated as a source of information, rather than support for decision-
48 making. Decisions were often made autonomously and sometimes conflicted with
49 parents' wishes. Participants reported no changes in health behaviours after testing. This
50 evidence highlights the need for a comprehensive, longitudinal counselling process with
51 appropriate timing and setting, which supports 'parent-to-offspring' risk communication
52 first and decision-making by young adults about presymptomatic testing and risk
53 management afterwards. Concluding, it is clear that counselling approaches for
54 presymptomatic testing may require modification both for young adults and their
55 parents.

56 **Key Words.**

57 Decision-making, genetic counselling, grounded theory, hereditary cancer, young
58 adults, presymptomatic genetic testing

59

60 **INTRODUCTION**

61 Presymptomatic genetic testing (PST) involves testing to determine if a person has
62 inherited a gene variant that causes a condition known to be present in the family,
63 before they exhibit any signs or symptoms of the condition. Those at risk of heritable
64 genetic disorders, including hereditary cancer syndromes¹ may be able to access PST to
65 determine their genetic status and potentially alter lifestyle choices or seek early
66 treatment for symptoms^{2,3}. Presymptomatic testing of minors (under the age of 18
67 years) in this situation is not usually recommended^{4,5}, although the age at which young
68 people should be able to undergo PST for adult-onset disorders is a matter of debate^{5,6}.
69 Key challenges that typically have to be faced during the transition from adolescence to
70 adulthood include marriage, completing education, beginning full-time employment and
71 becoming a parent: the impact of testing may affect, and be affected by, each of these
72 events.

73 A variety of psychosocial responses have been observed in those who have chosen to be
74 tested⁷. The appropriate age to offer PST is a matter of debate: it is suggested that
75 undergoing PST too early in life may increase the risk of unfavourable impact⁸⁻¹⁰. For
76 these reasons, individuals aged less than 18 years are not usually offered PST for adult-
77 onset disorders, the exceptions being if testing is considered to be in a child's best
78 interests⁴. Conversely, according to guidelines used in the United Kingdom (UK),
79 people aged 16 or 17 years are presumed to be capable of consenting to their own
80 medical treatment, and, in specific cases, children under 16 years who are adjudged to
81 fully understand what is involved in a proposed intervention will also have the capacity
82 to consent to that intervention¹¹: in other European countries adolescents have access to
83 medical treatment by law. In addition, it has been argued that young persons who are

84 considered as adults on the age-based criterion of 18 years are not all necessarily truly
85 autonomous⁹. There is no specific age when a person is able to give autonomous
86 consent, but it is important to consider psychological maturity⁹ that is cumulative with
87 age, life experience and cognitive development¹², while **maturity of judgement depends**
88 **upon responsibility, temperance, and perspective¹²**.

89 Prior to testing, young adults (YA) need to be aware of the potential risk to them of
90 hereditary cancer, and this is usually disclosed by their parents¹³⁻¹⁵. Prevalence and
91 experiences of parental communication of BRCA results to children under the age of 25
92 were described by Bradbury et al.¹⁶: 55% of parents (n=23/25) reported sharing family
93 history and/or genetic risk with at least one child. Their results indicate that the 42.9%
94 (n=18) of children in these families were learning of their potential genetic risk of
95 cancer before the age of 18 and 57% (n=24) between 18 and 25 years of age. It came to
96 light in that study that children of those with a BRCA **variant** learnt of their parent's
97 genetic test results many years before preventive interventions were indicated. In fact,
98 in a study of 273 women tested for hereditary breast and ovarian cancer **variant**,
99 Patenaude et al.¹³ noted that, although most children were told by their mother, the
100 child's age influenced the communication with offspring: they showed there was no
101 significant difference between numbers of minors (14 to 17 years, 85%) and YA (18 to
102 30 years, 92%) informed of the risk by their parents. Borry et al.⁴, in their paper on PST
103 in asymptomatic minors, concluded that minors, considering their age and degree of
104 maturity, are able to participate in decision-making and their opinions regarding PST
105 should be taken into consideration.

106 A systematic review¹⁷ on this topic indicated that many YA grew up with little or no
107 information concerning their genetic risk and that parents had exerted pressure during

108 the testing decision-making process. The experience of genetic counselling (GC) was
109 either reported as an opportunity for discussing problems or associated with feelings of
110 disempowerment. Moreover, emotional outcomes of disclosure did not correlate with
111 test results. However, none of the studies retrieved were conducted in Italy or other
112 South-European countries. To address this gap, we undertook a qualitative study based
113 on grounded theory to explore the psychosocial implications of PST for hereditary
114 cancer in Italian YA aged 18 to 30 years.

115 **MATERIALS AND METHODS**

116 This was a qualitative study in which we employed a grounded theory approach. This
117 approach was specifically chosen to explore the experiences of YA from their own
118 perspective, in this case the subjective meanings associated with being at risk for
119 hereditary cancer and their involvement with a health care technology and clinical
120 process for risk identification and reduction. This study received ethics approval both
121 from Plymouth University Faculty Research Ethics Committee (14/15-324), and St.
122 Orsola-Malpighi Hospital Ethical Board (132/2014/O/Oss).

123 In order to follow YA through the process of GC, from referral to follow-up, a
124 longitudinal study design was chosen. This enabled the authors to obtain data before
125 these were altered by the YA's contact with the genetic service, as well as providing the
126 opportunity to assess how perceived needs, expectations, and knowledge changed over
127 the period of contact. Each participant was interviewed on three separate occasions: one
128 month before GC, and two weeks and six months respectively after GC.

129 *Recruitment and participants*

130 All participants were recruited at the Genetics Unit of St.Orsola-Malpighi Hospital
131 (Bologna, Italy). Every new young consultand making an appointment for the cancer

132 genetics clinic was contacted before the consultation via telephone and invited to take
133 part in the study. Inclusion and exclusion criteria are presented in Figure 1. The process
134 of recruitment, interviews and data analysis was ongoing until data saturation¹⁸ was
135 reached and no new categories were emerging.

136 *Data collection*

137 Face-to-face interviews were organised with participants who responded to an invitation
138 to be involved in the study. All interviews were performed by LG (a genetic nurse with
139 training in counselling skills and five years of experience in GC), to ensure that the
140 participants were subject to a constant interviewer effect. Each interview began with
141 questions regarding demographic information. Later sections were designed to
142 understand the attitudes of YA, to evaluate their cancer perception and psychological
143 status and to explore the extent to which the parents' influence had been important. In
144 addition, questions were refined and amended over the course of the interviews to take
145 into account possible theories emerging from the data. The interviews were performed
146 with the participant only: any accompanying person was waiting outside. The
147 interviews were written in Italian (English version in Supplementary file). Data were
148 collected using a digital recording device and interviews were transcribed verbatim,
149 with names and other identifying material altered to ensure confidentiality.

150 *Data analysis*

151 Data were analysed using the grounded theory method¹⁸: each interview was analysed
152 as soon after transcription as possible. The software package NVivo, version 10 (QRS
153 international, Pty, Ltd) was used to help organise the data. The primary author listened
154 to the digital recordings and transcribed the interviews. Statements were subsequently
155 coded (open coding) from the transcribed material. All the interviews were translated

156 into English by LG and an expert translator and checked by DT to ensure accuracy of
157 translation and sense, and so improve rigour. The first 21 interviews were sent to other
158 co-authors (HS and LJ) to code independently to further ensure rigour and as there was
159 substantial agreement, the remaining interviews were coded by LG. The codes and
160 emerging categories derived by the co-authors were then compared to ensure
161 trustworthiness of the findings. Any disagreements were discussed until consensus was
162 reached. Finally, the data were further synthesised by grouping categories into major
163 themes to establish the relationships between data from all participants (axial coding).

164 **RESULTS**

165 Seventeen invitations were sent to potential participants: 14 (82.4%) accepted and were
166 interviewed (Figure 2). In total 42 interviews were conducted with 14 participants. The
167 participant characteristics are presented in Tables 1 and 2. Respondent ages ranged from
168 18 to 30, with a mean age of 25.3 years. The characteristics of the participants' parents
169 (based on information provided by the YA) are presented in a supplemental table.

170 Four major themes were identified. The pseudonym and interview number with that
171 participant (Int 1, 2 or 3) are included after each direct participant quote.

172 *Knowledge*

173 Many YA reported having grown up without awareness of or with misinformation about
174 the hereditary cancer running in their family. Following their first GC appointment,
175 some YA affirmed that as a result of the GC session their knowledge had improved:

176 *“Although the pathogenic gene contaminated the female organs, I thought my mom*
177 *could have transmitted the pathogenic gene to me and it could have contaminated every*
178 *single organ. At the beginning I was very confused.”* (Mario, age 26, Int 2)

179 Despite misinformation or lack of awareness, YA reported that the family history had
180 an important role in terms of their awareness and that it affected their feelings. *“Having*
181 *a family member diagnosed with cancer definitely makes you more aware of cancer.”*
182 (Donato, age 30, Int 3). Another important issue for participants was realizing the need
183 for surveillance. Some had not yet started any additional clinical surveillance that would
184 have been relevant for the familial condition. *“I want to prevent [...] I’ll do anything to*
185 *stay healthy [...] I want to live!”* (Barbara, age 29, Int 3). After GC, YA became aware
186 of the options for clinical screening, and the possibility of having more frequent
187 screening without undergoing PST: *“It was not required to proceed to the standard*
188 *routine of undergoing the exam, waiting for the results and then later entering the*
189 *screening; but you could choose to take up screening”* (Caterina, age 29, Int 2).
190 Nevertheless, one young woman thought that cancer could occur even if the variant was
191 not found and therefore she should have screening because of the family history: *“My*
192 *family history is that, despite the fact of having the syndrome or not.”* (Morena, age 25,
193 Int 1). Before GC, PST was described as ‘just a blood test’ by participants. Waiting for
194 the PST result was another point emphasised by YA. Some reported that was the only
195 thing they wanted to ask the genetic counsellor, for example, one young woman said:
196 *“At the end, I had only one question left and it was about the timing I had no*
197 *doubts ... but only lack of knowledge”* (Morena, age 25, Int 2). Also after the GC, the
198 PST was often perceived as ‘a need to wait for the result’. One young woman, who
199 experienced a pregnancy, had compared ‘the need to wait for the result’ with her
200 experience of finding out her baby’s gender. Although at first YA did not really know
201 what PST was, after GC they declared that they better understood what they were doing
202 or better understood the importance of undergoing PST. *“I truly understood (the*

203 *meaning of it all) only after dealing with counselling and questions they asked me”*
204 (Barbara, age 29, Int 3). At the same time, YA regarded the PST as a medical test like
205 any other. *“An exam like any other. [...] It was an ordinary blood sample.”* (Luca, age
206 24, Int 2). Once aware of the family genetic disorder, those who did not understand
207 what it really meant sought information online, while others did not want to use the
208 Internet as a source of research. Nevertheless, YA preferred not to speak about their
209 situation with friends. *“Then I sincerely don’t want to analyse my private life too much*
210 *with my friends.”* (Mario, age 26, Int 1). Almost all YA were informed of their family
211 genetic status by their mother. In cases where the mother was deceased, the person who
212 had been genetically tested in the family often informed the young adult.

213 *Genetic counselling process*

214 The experience of the GC process was explored and YA explained their motivations to
215 have it, their expectations and experience of it. Undergoing GC was motivated by
216 curiosity, a need for information, and to obtain certainty. Others focused their attention
217 on undergoing GC to help prevent cancer. The decision to undergo GC was not always
218 specifically discussed with parents, but YA knew that their relatives had consulted
219 medical professionals and wished to follow a similar pathway. Nevertheless, four
220 participants underwent GC purely for themselves, for example Mario decided to go
221 through GC: *“For a more serene future.”* (Mario, age 26, Int 1). One of the two YA
222 with children underwent GC because of anxiety about her daughter, while some
223 participants underwent GC to understand the risk to their future children.

224 The majority of YA interviewed had no expectations about GC, mostly because they
225 lacked knowledge about it. However, they still expected a blood test, as something that
226 genetic counsellors suggested, something they had to do, and something that would be

227 uncomfortable. *“Counselling was the prelude of the genetic test [...] I didn’t think I*
228 *could have said ‘no’ at the end as well as any other person. [...] I thought it was a*
229 *required step”* (Morena, age 25, Int 2). Some YA perceived GC/PST as a ‘*need to wait*
230 *for the result’*, and they were therefore surprised to have the blood sample taken at the
231 first consultation. *“I honestly didn’t expect to be tested during the first counselling.”*
232 (Barbara, age 29, Int 2). Young adults interviewed reported GC had helped them,
233 through the process of discussion with the counsellor. Some positive feelings were
234 expressed about genetic counsellors, such as the perception of being understood and that
235 the counsellor was the person who explained the meaning of testing. Many YA reported
236 that they had not expected to have a choice. They had assumed that, in agreeing to
237 undergo the GC process, they would have a PST and they were surprised when they
238 realized they make a testing decision. *“At the end, they asked me if I wanted to do this*
239 *thing. I thought counselling ended with the genetic test, instead it didn’t! It was the idea*
240 *I had for months!”* (Eleonora, age 30, Int 2). *All the YA were offered, and underwent,*
241 *PST at the first GC session except one, who was offered a second pre-test session. She*
242 *declared she felt more aware of the implications of the test when she underwent it*
243 *during the second session: “With hindsight I think the first time I’d have done it*
244 *unconsciously. [...] today, I’m more conscious about what I’m doing.”* (Paola, age 25,
245 Int 2). Even if they had already made a clear choice to undergo the PST before the
246 consultation, some expressed a desire to have the genetic counsellor give an opinion to
247 guide them.

248 *Decision-making for testing or not*

249 Although theoretically, making an autonomous choice to undergo PST is a fundamental
250 requirement of the process of GC, some young family members were subject to pressure

251 from their parents to be tested. As a consequence of parental pressure, some YA
252 reported that they underwent PST for the sake of a parent/relative. *“Honestly, because*
253 *my mother told me and she did it first ... I'm doing it as a favor to her.”* (Luca, age 24,
254 Int 1). However, differences emerged in the extent of parental involvement in the
255 decision-making process. In some cases, the decision to have a PST was made
256 autonomously but was congruent with the relatives’ point of view. *“I called to have an*
257 *appointment under pressure from my mother ... I'd have done it sooner or later.*
258 *...although I would have chosen to wait a bit more.”* (Angelica, age 24, Int 1). On the
259 other hand, the decision was sometimes at odds with the parent’s opinion. *“She*
260 *(mother) has always been very uncertain whether to get me to do the project. She said:*
261 *‘You have to think more deeply about it, the result doesn’t change’.*” (Morena, age 25,
262 Int1). The participants’ decision-making process occurred before the first GC session:
263 no participant reported having GC to help facilitate their decision about testing.
264 However, it was not clear whose idea it was to undergo PST. Some of them tried to
265 align the decision to have counselling with their perception of the appropriate time to
266 start clinical surveillance. **The majority attended the GC session alone, however, even if**
267 **the participant attended alone, the counselling session was often arranged by the**
268 **participant’s mother, especially for young men. No differences emerged between those**
269 **whose mothers had booked and those who booked themselves. Nevertheless, a young**
270 **woman who had decided to bring her mother with her reported that “Having her there**
271 **made me experience the counselling as way more touching”** (Morena, age 25, Int 2).
272 The majority of YA decided not to share the decision to undergo PST with their friends.
273 Others decided to share it only with close friends because they felt that other people
274 would not understand the complexities of the situation. As Barbara (age 29, Int 1)

275 described: “None of my friends knows (what I’m doing) because I think these are very
276 personal things and, knowing my friends, I’m afraid that some of them might think bad
277 (of me) and then I would feel bad”. Looking back on their experience of PST, three
278 participants expressed a desire for something different from what they had experienced.
279 While Barbara suggested a YA support group to discuss experiences, share ideas, and
280 provide emotional support, others proposed having more professional psychological
281 support.

282 *Dealing with the result*

283 Some participants perceived PST as a source of tension, mostly before they underwent
284 GC. As Dario (age 20, Int 3) described: “At the beginning, it is normal to feel a little bit
285 scared or worried because it is something unknown ... but when everything is explained
286 one calms down”. Some YA expected that the PST result would be negative. Others,
287 who believed before testing that they would be variant-positive, felt relieved when the
288 test had a different outcome. As Barbara (age 29, Int 1) described: “If I didn’t have the
289 gene ... breathe”. However, the PST result was perceived by YA as useful in helping
290 them to plan their lives. Conversely, others did not think that they would change their
291 behaviour based on the possible result. However, when they considered how they might
292 react, the majority affirmed that they did not know.

293 Once aware of their test result, none of those interviewed reported a catastrophic
294 emotional response: emotions of relief, happiness and fear were generally reported.
295 Accordingly, participants with negative PST results described themselves and their
296 parents as happy to have this knowledge. Regardless of the result, some YA felt they
297 had matured as a result of their testing experience. Moreover, once they had received
298 the result, they recommended that their relatives (e.g. siblings) undergo PST as well.

299 Only Morena specifically recommended GC to her relatives. Changes in behaviour were
300 not generally reported in either **variant**-positive or **variant**-negative YA, however, a
301 young woman who was **variant**-positive started to pay more attention to her body and
302 possible symptoms.

303 Young women who were **variant**-positive, started their surveillance and one of them
304 described herself '*having butterflies*' (Barbara, **age 29**, Int 3) after her first screening,
305 nervous about her first ultrasound outcome. Fortunately, it was normal and she felt
306 relieved, but she underlined that the relief would last '*until the next follow-up visit*'.

307 **DISCUSSION**

308 **The aim of this study was to** investigate the experience of PST in Italian YA aged 18-30
309 years. **The choice of this range of age was made on the basis of the specific Italian**
310 **context. In Italy, the age at which YA leave the parental home is very high when**
311 **compared to other countries^{19,20}. It is clear that the activities of young adulthood, e.g.**
312 **forming partnerships and becoming parents, occur later than in other cultures²⁰ and this**
313 **could affect their PST decision-making.**

314 The results show that participants grew up with little or no information about their
315 genetic risk and they usually became aware of their risk less than one year before
316 testing. This is in contrast with findings emerging from the papers reviewed in the
317 systematic review¹⁷ where YA were informed several years before testing or clinical
318 actions could be undertaken²¹⁻²³. **Considering the Italian context, this may be because of**
319 **the delay of YA's development into mature adulthood.** At the time of the final
320 interview, young adults were consciously, as well as unconsciously, developing
321 strategies to cope with the experience they were facing. There was a dynamic
322 relationship between the decision-making process and their autonomous choice: YA

323 arrived at the decision-making process because of previous knowledge, disclosed by
324 one or both parents. Consistent with this finding, a meta-synthesis of the family
325 communication between children and their parents about inherited genetic conditions
326 conducted by Metcalfe et al.²⁴ showed that parents were primarily responsible for
327 discussing genetic information with their children. Although there was a desire by
328 parents to tell their children about their potential genetic risk before others told them²⁴,
329 parents also stressed delaying the disclosure or choosing the right time to talk²⁵. No
330 differences emerged between participants who underwent PST when they aged less than
331 24 years and those who were older, whereas Hamilton²⁶ reported that older YA were
332 more likely than younger ones to decide autonomously to have PST. Young adults are
333 normally at a stage of life in which they are acquiring knowledge about themselves and
334 the world around them^{27,28}. They may or may not be sufficiently mature, or have a
335 realistic set of expectations about what genetic information will allow them to do, or
336 even the health insurance to support risk management decision-making^{12,29}. They may
337 or may not fully understand the science behind PST related cancer risk, gene
338 penetrance, or prevention.

339 In this study, at the start of the GC process participants had often not understood that
340 their choices had serious implications. Instead, as Lindenmeyer et al.³⁰ underlined,
341 participants did not choose to undergo PST separate from the collective concerns and
342 desires of their families. Parents may exert pressure on YA children to complete PST³¹,
343 however no participants reported the same behaviour as their parents in terms of risk
344 management decisions (e.g. surgery rather than screening).

345 Concerning the impact of test results, overall, our findings do not support a substantial
346 risk of adverse emotional outcome in **variant** carriers, which is in agreement with

347 previous findings³². In contrast, being variant-positive for Huntington disease may
348 influence a YA's education, career, relationships and family planning³³. This may be
349 because there is no preventive treatment available at present for that condition, or that
350 the condition is perceived to have much greater impact on functioning throughout life.

351 Overall, although our results may not be generalizable because of a lack of data from
352 South-European countries, differences with other countries emerged. Further study in
353 the Mediterranean area may be needed to clarify if these differences are peculiar to the
354 Italian population or may be generalizable to other countries of this area.

355 *Strengths and limitations*

356 An identifiable strength of this study was the method chosen, which provided an
357 effective framework for key themes to emerge from the data. Moreover, because of the
358 longitudinal design we have been able to ascertain the views of young adults
359 considering testing both prospectively and retrospectively. A limitation of this study is
360 that we only identified YA who decided to undergo PST, as we were unable to recruit
361 YA who decided not to be tested. Additionally, we were unable to affirm that our results
362 are unique for the age group studied, the comparison with older age groups was not
363 possible as it falls outside the aim of the present study.

364 **CONCLUSION**

365 The findings of this study indicate a need for further guidance on PST in these
366 populations: it is important for health professionals to understand how much the YA
367 involved are really aware of the implications before and after they have been tested. It is
368 therefore important to publicise the supportive and educational role of genetic services.
369 Moreover, appropriate communication of genetic risk information by parents to their
370 children is highly desirable, since it has been shown to have long-term consequences²⁴.

371 To achieve this, health professionals could have a role in both supporting parents and
372 YA, as their involvement in the parents' decision to communicate genetic risk to young
373 family members was found to be limited^{16,17,34,35}. Although this may be partly due to the
374 parents' wish to undertake this task alone, it is reported that some parents desired health
375 professionals to be available in a supporting role, but found this was limited^{24,36}. This
376 evidence highlights the need for a comprehensive, longitudinal counselling process with
377 appropriate timing and setting, which supports 'parent-to-offspring' risk communication
378 first and YA's decision-making about PST and risk management afterwards. In
379 conclusion, it is clear that GC approaches to this population may require modification
380 both for YA and their parents. Further analysis is required to determine how YA and
381 their parents interpret PST, how they experience GC, and the influence that parents have
382 on YA's decisions after the disclosure of the positive test result to inform GC practice in
383 this client group.

384 **ACKNOWLEDGEMENTS**

385 We would like to express our gratitude to the YA who participated in this study. We
386 also acknowledge the Medical Genetics Unit staff of Bologna for identifying and
387 suggesting potential participants and Dr. Annalisa Spinelli for helping us to translate the
388 interviews into English. LG was supported by the Grant from Regione Emilia-Romagna
389 "Diagnostics advances in hereditary breast cancer (DIANE)" (PRUa1GR-2012-001).

390 **CONFLICTING INTEREST**

391 The authors have no conflicts of interest.

392 **REFERENCES**

- 393 1 Evans JP, Skrzynia C, Burke W. The complexities of predictive genetic testing.
394 *BMJ* 2001; **322**: 1052–6.

- 395 2 Beery TA, Williams JK. Risk reduction and health promotion behaviors
396 following genetic testing for adult-onset disorders. *Genet Test* 2007; **11**: 111–
397 123.
- 398 3 Quaid KA, Sims SL, Swenson MM, Harrison JM, Moskowitz C, Stepanov N *et*
399 *al.* Living at risk: concealing risk and preserving hope in Huntington disease. *J*
400 *Genet Couns* 2008; **17**: 117–28.
- 401 4 Borry P, Evers-Kiebooms G, Cornel MC, Clarke A, Dierickx K. Genetic testing
402 in asymptomatic minors: background considerations towards ESHG
403 Recommendations. *Eur J Hum Genet* 2009; **17**: 711–719.
- 404 5 MacLeod R, Tibben A, Frontali M, Evers-Kiebooms G, Jones A, Martinez-
405 Descales A *et al.* Recommendations for the predictive genetic test in
406 Huntington’s disease. *Clin Genet* 2013; **83**: 221–231.
- 407 6 Borry P, Evers-Kiebooms G, Cornel MC, Clarke A, Dierickx K. Genetic testing
408 in asymptomatic minorsBackground considerations towards ESHG
409 Recommendations. *Eur J Hum Genet* 2009; **17**: 711–719.
- 410 7 Baig SS, Strong M, Rosser E, Taverner N V, Glew R, Miedzybrodzka Z *et al.* 22
411 Years of predictive testing for Huntington’s disease: the experience of the UK
412 Huntington’s Prediction Consortium. *Eur J Hum Genet* 2016; **24**: 1396–1402.
- 413 8 Borry P, Stultiens L, Nys H, Cassiman J-J, Dierickx K. Presymptomatic and
414 predictive genetic testing in minors: a systematic review of guidelines and
415 position papers. *Clin Genet* 2006; **70**: 374–381.
- 416 9 Richards FH. Predictive genetic testing of adolescents for Huntington disease: A
417 question of autonomy and harm. *Am J Med Genet Part A* 2008; **146A**: 2443–
418 2446.
- 419 10 Duncan RE, Gillam L, Savulescu J, Williamson R, Rogers JG, Delatycki MB.
420 Reply to Richards: ‘Predictive genetic testing of adolescents for Huntington
421 disease: A question of autonomy and harm’. *Am J Med Genet Part A* 2008;
422 **146A**: 2447–2448.

- 423 11 Department of Health. *Reference guide to consent for examination or treatment*,
424 *second edition 2009*. London, 2009.
- 425 12 Steinberg L, Cauffman E. Maturity of judgment in adolescence: Psychosocial
426 factors in adolescent decision making. *Law Hum Behav* 1996; **20**: 249–272.
- 427 13 Patenaude AF, Dorval M, DiGianni LS, Schneider KA, Chittenden A, Garber JE.
428 Sharing BRCA1/2 test results with first-degree relatives: Factors predicting who
429 women tell. *J Clin Oncol* 2006; **24**: 700–706.
- 430 14 Bradbury AR, Patrick-Miller L, Egleston BL, Olopade OI, Daly MB, Moore CW
431 *et al*. When parents disclose BRCA1/2 test results: Their communication and
432 perceptions of offspring response. *Cancer* 2012; **118**: 3417–3425.
- 433 15 Van Der Meer LB, Van Duijn E, Wolterbeek R, Tibben A. Adverse childhood
434 experiences of persons at risk for Huntington’s disease or BRCA1/2 hereditary
435 breast/ovarian cancer. *Clin Genet* 2012; **81**: 18–23.
- 436 16 Bradbury AR, Dignam JJ, Ibe CN, Auh SL, Hlubocky FJ, Cummings SA *et al*.
437 How often do BRCA mutation carriers tell their young children of the family’s
438 risk for cancer? A study of parental disclosure of BRCA mutations to minors and
439 young adults. *J Clin Oncol Off J Am Soc Clin Oncol* 2007; **25**: 3705–3711.
- 440 17 Godino L, Turchetti D, Jackson L, Hennessy C, Skirton H. Impact of
441 presymptomatic genetic testing on young adults: a systematic review. *Eur J Hum*
442 *Genet* 2016; **24**: 496–503.
- 443 18 Corbin JM, Strauss AL. *Basics of qualitative research : techniques and*
444 *procedures for developing grounded theory*. 2014.
- 445 19 Ferrari G, Rosina A, Sironi E. Beyond good intentions: the decision-making
446 process of leaving the family of origin in Italy. *Dondena Work Pap* 2014; **60**: 1–
447 23.
- 448 20 Istat. *Giovani.Stat*. 2016.<http://dati-giovani.istat.it/Index.aspx> (accessed 10
449 Dec2016).

- 450 21 Duncan RE, Gillam L, Savulescu J, Williamson R, Rogers JG, Delatyckil MB.
451 'Holding your breath': Interviews with young people who have undergone
452 predictive genetic testing for Huntington disease. *Am J Med Genet Part A* 2007;
453 **143A**: 1984–1989.
- 454 22 Patenaude AF, Tung N, Ryan PD, Ellisen LW, Hewitt L, Schneider KA *et al.*
455 Young adult daughters of BRCA1/2 positive mothers: What do they know about
456 hereditary cancer and how much do they worry? *Psychooncology* 2013; **22**:
457 2024–2031.
- 458 23 Hoskins LM, Werner-Lin A, Greene MH. In their own words: treating very
459 young BRCA1/2 mutation-positive women with care and caution. *PLoS One*
460 2014; **9**: e87696.
- 461 24 Metcalfe A, Coad J, Plumridge GM, Gill P, Farndon P. Family communication
462 between children and their parents about inherited genetic conditions: a meta-
463 synthesis of the research. *Eur J Hum Genet* 2008; **16**: 1193–1200.
- 464 25 Metcalfe A, Plumridge G, Coad J, Shanks A, Gill P. Parents' and children's
465 communication about genetic risk: a qualitative study, learning from families'
466 experiences. *Eur J Hum Genet* 2011; **19**: 640–646.
- 467 26 Hamilton R. Being young, female, and BRCA positive. *Am J Nurs* 2012; **112**: 26.
- 468 27 Arnett JJ, Jensen J. Emerging adulthood: A theory of development from the late
469 teens through the twenties. *Am Psychol* 2000; **55**: 469–480.
- 470 28 Arnett JJ, Tanner JL. *Emerging adults in America: Coming of age in the 21st*
471 *century*. American Psychological Association: Washington, DC, 2006.
- 472 29 Richards FH. Maturity of judgement in decision making for predictive testing for
473 nontreatable adult-onset neurogenetic conditions: a case against predictive testing
474 of minors. *Clin Genet* 2006; **70**: 396–401.
- 475 30 Lindenmeyer A, Griffiths F, Hodson J. 'The family is part of the treatment
476 really': a qualitative exploration of collective health narratives in families. *Health*
477 *(London)* 2011; **15**: 401–415.

- 478 31 Hoskins LM, Roy KM, Greene MH. Toward a new understanding of risk
479 perception among young female BRCA1/2 ‘previvors’. *Fam Syst Heal* 2012; **30**:
480 32–46.
- 481 32 Broadstock M, Michie S, Marteau T. Psychological consequences of predictive
482 genetic testing: a systematic review. *Eur J Hum Genet* 2000; **8**: 731–738.
- 483 33 Gong P, Fanos JH, Korty L, Siskind CE, Hanson-Kahn AK. Impact of
484 Huntington Disease Gene-Positive Status on Pre-Symptomatic Young Adults and
485 Recommendations for Genetic Counselors. *J Genet Couns* 2016.
486 doi:10.1007/s10897-016-9951-z.
- 487 34 Rew L, Mackert M, Bonevac D. A Systematic Review of Literature About the
488 Genetic Testing of Adolescents. *J Spec Pediatr Nurs* 2009; **14**: 284–294.
- 489 35 Werner-Lin A, Ratner R, Hoskins LM, Lieber C. A survey of genetic counselors
490 about the needs of 18-25 year olds from families with hereditary breast and
491 ovarian cancer syndrome. *J Genet Couns* 2015; **24**: 78–87.
- 492 36 Gaff CL, Lynch E, Spencer L. Predictive testing of eighteen year olds:
493 counseling challenges. *J Genet Couns* 2006; **15**: 245–251.
- 494

495 **TITLES AND LEGENDS TO FIGURES**

496 **Figure 1:** INCLUSION AND EXCLUSION CRITERIA

497 **Figure 2:** THE RECRUITMENT PROCESS

498 **Table 1:** DESCRIPTION OF PARTICIPANTS' CHARACTERISTICS

499 **Table 2:** DESCRIPTION OF THE SOCIAL BACKGROUND OF EACH PARTICIPANT

500

FIGURE 1: INCLUSION AND EXCLUSION CRITERIA

Participants were invited to take part in the study if they were:

- aged 18-30 years
- without personal history of cancer
- members of families with a hereditary cancer predisposition
- able to give informed consent, and
- able to speak Italian or English fluently.

Young adults were excluded from the study if they were:

- clients counselled by the principal researcher
- unable to provide informed consent, for example due to mental incapacity or active psychotic illness.

FIGURE 2: THE RECRUITMENT PROCESS

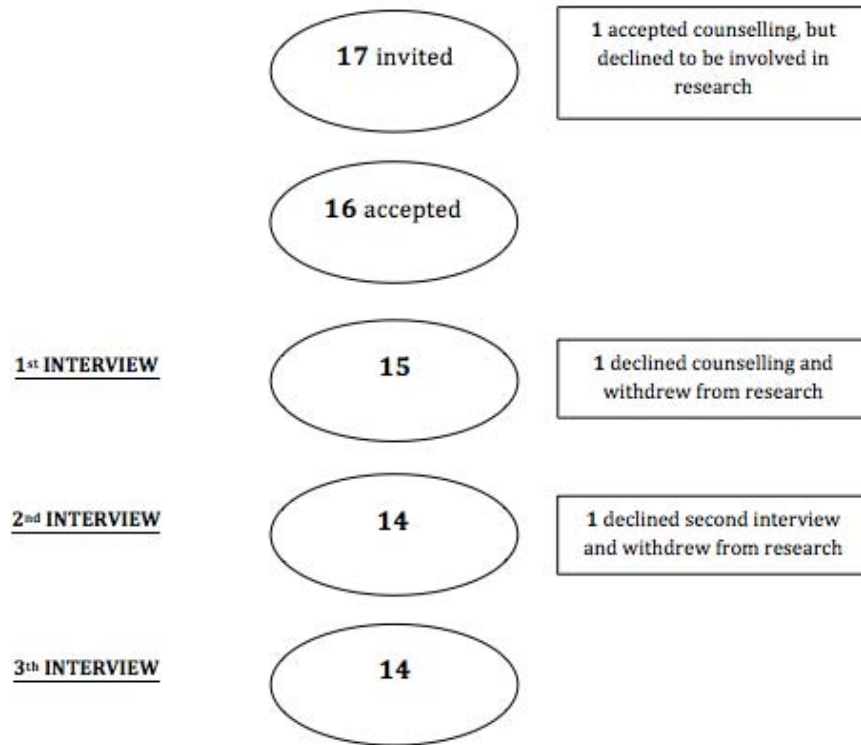


TABLE 1: DESCRIPTION OF PARTICIPANT CHARACTERISTICS

GENDER N(%)	
MALE	6 (42.9%)
FEMALE	8 (57.1%)
AGE AT INTERVIEW (YEARS)	
MEAN±SD	25.3±3.6
COUNTRY OF BIRTH N(%)	
ITALY	13 (92.9%)
POLAND	1 (7.1%)
MOTHER'S LANGUAGE N(%)	
ITALIAN	13 (92.9%)
POLISH	1 (7.1%)
CONDITION TESTED FOR N(%)	
BRCA1	8 (57.2%)
BRCA2	5 (35.7%)
MLH1	1 (7.1%)
AGE AT TEST (YEARS)	
MEAN±SD	25.3±3.6
RANGE	18-30
RESULT N(%)	
POSITIVE (FOR MUTATION)	4 (28.6%)
NEGATIVE (FOR MUTATION)	10 (71.4%)
EDUCATION N(%)	
MIDDLE SCHOOL QUALIFICATION	1 (7.1%)
HIGH SCHOOL DIPLOMA	7 (50.0%)
UNIVERSITY DEGREE	5 (35.8%)
POST-GRADUATE DEGREE	1 (7.1%)
DAILY WORK N(%)	
STUDENT	5 (35.8%)
WORKER	3 (21.4%)
EMPLOYEE	3 (21.4%)
BUSINESS OWNER	1 (7.1%)
UNEMPLOYED	2 (14.3%)
MARITAL STATUS N(%)	
MARRIED	1 (7.1%)
SINGLE	12 (85.8%)

LIVING TOGETHER	1 (7.1%)
HAVING CHILDREN N(%)	
NO	12 (85.7%)
YES	2 (14.3%)

TABLE 2: DESCRIPTION OF THE SOCIAL BACKGROUND OF EACH PARTICIPANT

ID	AGE AT INTERVIEW (YEARS)	GENERAL INFORMATION	CARRIER PARENT	COMMUNICATION OF FAMILIAL MUTATION	INTERVIEW DURATION (MIN)
Donato	30	<p>His mother was diagnosed with ovarian cancer when he was 26 years old. One maternal aunt had breast cancer some years ago. His mother was the first person in the family to have genetic testing and she discovered her result one year ago.</p> <p>He lives in various countries around the world because of his work.</p> <p>The interviews were very difficult to arrange due to challenges in communication and making time. In fact the last interview was conducted by email.</p>	Mother	Mother	<p>Int.1 – 14.21</p> <p>Int.2 – email</p> <p>Int.3 - email</p>
Barbara	29	<p>Her mother was diagnosed with ovarian cancer when she was 26 years. On her mother’s side, her grandmother and one aunt also had breast cancer. Her mother was the first person in the family to have genetic testing and she discovered her result two years ago. Both grandmother and aunt had genetic testing and both have BRCA mutations.</p> <p>She has lived in Italy since she was 20 years old.</p> <p>She gave the impression of being a very strong young woman, however she was accompanied by her mother at both interviews and counselling sessions.</p>	Mother	Mother	<p>Int.1 – 38.51</p> <p>Int.2 – 10.54</p> <p>Int.3 – 44.35</p>
Morena	25	<p>Her mother was diagnosed with colon cancer when she was 8 years old, and with endometrial cancer when she was 19 years old. On her mother’s side, her grandfather had colon cancer and his mother was diagnosed with gynaecological cancer. Her mother was the first person in the family to have genetic testing and she discovered her result six years ago.</p> <p>She was accompanied by her mother at counselling sessions. She also texted me to remind me about her interviews.</p>	Mother	Mother	<p>Int.1 – 41.22</p> <p>Int.2 – 43.25</p> <p>Int.3 – 42.01</p>
Mario	26	<p>His mother was diagnosed with breast cancer when he was 13 years old. In the same period, a maternal aunt had breast cancer. Another maternal aunt had</p>	Mother	Mother	<p>Int.1 – 22.53</p> <p>Int.2 – 24.23</p>

		breast cancer when he was 20 years old. His maternal grandmother died because of ovarian cancer when he was 22 years old. His grandmother was the first person in the family to have genetic testing and his mother discovered her genetic status one year ago.			Int.3 – 12.31
		He lives in a small city in the south of Italy.			
		He texted me to remind me about his interviews, however he was accompanied by his mother and his maternal uncle at the counselling session. His result was collected by his maternal uncle.			
Angelica	24	Her mother was diagnosed with breast cancer when she was 22 years old. Maternal grandmother died because of breast cancer. Her mother was the first person in the family to have genetic testing and his mother discovered her genetic status one year ago.	Mother	Mother	Int.1 – 26.18 Int.2 – 30.48 Int.3 – 18.56
		She came alone to both interviews and counselling, however she forgot both her first counselling session and our second interview. She only remembered after receiving an appointment reminder.			
Paola	25	Two paternal aunts were diagnosed with breast cancer and another paternal aunt had ovarian cancer. Grandmother died because of ovarian cancer. Recently her father discovered his genetic status.	Father	Father and aunts (father's side)	Int.1 – 19.47 Int.2 – 20.28 Int.3 – 21.01
		She came alone to both interviews and counselling sessions.			
Eleonora	30	Her mother died because of breast cancer, as did two maternal aunts. Her grandmother was the first person in the family to have genetic testing and she discovered her genetic status one years ago.	Mother (?)	Cousin	Int.1 – 22.55 Int.2 – 28.41 Int.3 – 24.21
		She texted me to remind me about her interviews, however she was accompanied by her father both at interviews and at the counselling sessions. Although he was with her during the counselling, she never mentioned this.			
Luca	24	His mother was diagnosed with breast cancer last year. His maternal grandmother was diagnosed with ovarian cancer and breast cancer when he was 20 years. His grandmother was the first person in the family to have genetic testing and his mother discovered her genetic status one year ago.	Mother	Mother	Int.1 – 10.01 Int.2 – 11.56 Int.3 – 12.35

		He was accompanied by a friend at the counselling sessions.			
		The interviews were very difficult to arrange in terms of communication.			
Caterina	29	Her mother was diagnosed with ovarian cancer when she was 27 years. On her mother's side, two aunts had breast cancer and grandmother had ovarian cancer. One aunt was the first person in the family to have genetic testing and mother discovered her genetic status two years ago.	Mother	Mother	Int.1 – 25.40 Int.2 – 24.07 Int.3 – 26.09
		She came alone to both interviews and counselling sessions.			
Emma	27	Her mother was diagnosed with breast cancer when she was 25 years. Maternal grandmother died because of breast cancer. Her mother was the first person in the family to have genetic testing and his mother discovered her genetic status one year ago. Some months before, her sister (Angelina here) was tested.	Mother	Mother	Int.1 – 16.46 Int.2 – 14.50 Int.3 – 17.35
		She came alone to both interviews and counselling.			
Patrizia	23	Her mother was diagnosed with breast cancer when she was 6 years old and with contralateral breast cancer when she was 20 years old. Her maternal aunt had breast cancer when she was 21. Her mother was the first person in the family who had genetic testing and his mother discovered her genetic status two years ago.	Mother	Mother	Int.1 – 17.51 Int.2 – 10.51 Int.3 – 24.20
		She was accompanied by maternal aunt both at interviews and counselling sessions.			
Dario	20	His mother was diagnosed with breast cancer when he was 2 years and with contralateral breast cancer when he was 17 years old. Both his maternal aunt and grandmother had breast cancer. His mother was the first person in the family to have genetic testing and his mother discovered her genetic status one year ago.	Mother	Mother	Int.1 – 20.29 Int.2 – 17.35 Int.3 – 21.53
		He was accompanied by his brother both at first interview and first counselling session. He came alone to the post-test counselling and his brother delegated him to collect the brother's genetic test result (in Italy this is not routine, but sometimes happens).			

Matteo	18	<p>His mother was diagnosed with breast cancer when he was 17 years old. His mother was the first person in the family to have genetic testing and his mother discovered her genetic status one year ago.</p> <p>He was accompanied by a friend both at the interviews and at counselling sessions.</p>	Mother	Mother	<p>Int.1 – 38.51</p> <p>Int.2 – 10.54</p> <p>Int.3 – 44.35</p>
Saverio	24	<p>Two maternal aunts were diagnosed respectively with breast cancer and ovarian cancer. Recently his mother discovered her genetic status.</p> <p>The interviews were very difficult to arrange in terms of communication and time.</p>	Mother	Mother	<p>Int.1 – 10.51</p> <p>Int.2 – 09.55</p> <p>Int.3 – 11.01</p>