**BABEȘ-BOLYAI UNIVERSITY**
**FACULTY OF PSYCHOLOGY AND EDUCATIONAL SCIENCES**
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# Extended Summary of PhD. Thesis

## Genetic counselling for young people: principles and practice

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# Contents

## Abstract

Genetic counselling is a well-established and empirically supported service, with precise clinical guidelines, protocols and ethical frameworks. Genetic counselling is traditionally offered to adult patients. Children and adolescents are increasingly starting to access this service and several empirical studies, guidelines and recommendations on this topic have been published, but the literature is scarce and heterogenous. The main aim of this thesis is to investigate genetic counselling for young people and their families, by integrating the perspectives and needs from all stakeholders involved (e.g. children, adolescents, parents, health professionals) and by documenting and implementing an evidence-based counselling intervention. The first study is aimed at reviewing the current state of published literature on genetic counselling for young people. The second and third studies investigate the needs (i.e. medical, informational, communicational and psychosocial) and the emotional distress (e.g. levels of stigma, burden, well-being) experienced by families with complex health needs. The fourth study explores current practices and needs of genetic counsellors working with children and adolescents. The fifth and final study is designed to investigate the impact of genetic counselling for parents of children and adolescents with psychiatric conditions.

The research in this thesis adds several important contributions to the field: (1) theoretical, through the systematic review, (2) practical, by better understanding the needs of all stakeholders involved (e.g. young people, parents, genetic counsellors), and (3) clinical, by documenting the efficacy of psychiatric genetic counselling for parents who have children diagnosed with psychiatric conditions.

The thesis includes four chapters: (1) theoretical framework of genetic counselling, with particular focus on genetic counselling for children and adolescents and on psychiatric genetic

counselling; (2) methodology of the thesis; (3) original research; and (4) results and contributions of conducted research.

**Key words:** genetic counselling, psychiatric genetic counselling, young people, parents, genetic counsellors, psychiatric conditions, genetic conditions, complex needs, current practices, pilot intervention

# Chapter I. Theoretical framework

## I. Introduction

### 1. Genetic counselling

Genetic counselling is "the process of helping people understand and adapt to the medical, psychological, and familial implications of the genetic contributions to disease. This process integrates: (a) interpretation of family and medical histories to assess the chance of disease occurrence or recurrence; (b) education about inheritance, testing, management, prevention, resources and research; and (c) counselling to promote informed choices and adaptation to the risk or condition" (Resta et al., 2006, p.77).

#### *1.1. Evolution of genetic counselling*

Originally, the main focus of practice and research in genetic counselling was mainly reproduction issues, particularly on preventing developmental disorders. In the 1970s, research and practice looked increasingly more into the importance and complexity of the psychological processes involved in genetic counselling, such as decision making, emotional distress and reproductive behaviours/intentions (Resta, 2019). In the 1980s, important questions about the process of genetic counselling were asked: what are the needs of the patients, how do they perceive risk assessment, how does genetic information change their deliberation and choice process, what makes genetic counselling effective, what outcomes should be measured, how does genetic counselling impact patients, etc. (Resta, 2019). In the 1990s, genetic counselling became a separate specialty from medical genetics and expanded its applicability in other medical areas, like oncology and cardiology (Resta, 2019).

### *1.2. Models of practice in genetic counselling*

Several conceptual frameworks and models of practice have been developed based on theoretical models from medicine, mental health and education (McCarthy Veach et al., 2007). Kessler was the first to articulate two models of practice for genetic counselling: the teaching model and the counselling model (Kessler, 1997). The teaching model approaches genetic counselling as being mostly focused on presenting information and educating the patient about genetics and medicine, without a particular emphasis on psychosocial aspects. The counselling model gives the patient a more central and active role, by taking into consideration the patients' needs, questions, values, decisional processes and personal goals.

The Reciprocal Engagement Model (REM, McCarthy Veach et al., 2007), more recently published, integrates both medical and psychological aspects, as well as the therapeutic relationship as key ingredients of the genetic counselling process.

### *1.3. Professional and ethical principles in genetic counselling*

Several codes of practice and conduct have been developed and revised along the years (Senter et al., 2017). The main ethical principles of the profession are autonomy, non-directiveness, beneficence, non-maleficence, justice and client-centred care (Cohen, 2020; National Society of Genetic Counselors, 2017). The National Society of Genetic Counselors has established that genetic counsellors have responsibilities towards and have to manage their relationship with (1) their patients, (2) themselves, (3) their colleagues and (4) society, while at the same time exercising "fidelity, veracity, integrity, dignity and accountability" in their practice (National Society of Genetic Counsellors, 2017).

### *1.4. Process of genetic counselling*

People may attend genetic counselling for several reasons (Process of Genetic Counselling GL02 -Human Genetics Society of AustralAsia, 2012):

- The patient is at risk/has a family history of a genetic condition
- The patient is at risk/has a family history of being a carrier for a possible genetic condition
- The patient has a pregnancy with foetal abnormalities or has a history of births with developmental problems and/or intellectual disabilities and wants to know more about future pregnancies
- The patient requires information/interpretation of genetic test results (i.e. newborn screening result, prenatal screening results, direct to consumer testing)
- The client/family has a history of severe mental health problems

### *1.5. Education and training in genetic counselling*

Education includes information about human genetics, medical genetics and clinical aspects of genetic disease, genomic information, counselling skills, knowledge about working in various clinical areas and psychosocial issues, ethical, social and law principles.

Currently, genetic counsellors can specialise and work in a more general setting (e.g. rare genetic diseases, chromosomal anomalies) or in a more specific area of genetics like cardiac genetic counselling, cancer genetic counselling, paediatric genetic counselling, psychiatric genetic counselling etc. (Abacan et al., 2018; Genetic Alliance, n.d.).

### *1.6.* E*fficacy of genetic counselling*

Counselling brings long-term benefits to both patients and family members (Moldovan et al., 2017), it improves knowledge, perceived risk, psychological well-being (e.g. anxiety, distress, empowerment, self-efficacy) and patient satisfaction (Athens et al., 2017; Braithwaite et al., 2006; Bracke et al., 2020; Meiser & Halliday, 2002; Moldovan et al., 2017). Comparable results have been found between in person or other delivery settings (e.g. on-line, telephonic) (Bracke et al., 2020). Nonetheless, there are several gaps in the literature (e.g. efficacy of genetic counselling for children and adolescents) which need to be researched in order to continue to deliver evidence-based practices.

## 2. Genetic counselling for children and adolescents

Genetic counselling for children and adolescents has become a topic of growing interest in both clinical practice and research. Whilst early studies were mainly focused on specific genetic disorders and explored primarily parents' perspectives (see Dubowitz, 1975; Fraser, 1958; Lessick et al., 1981; Markova et al., 1984; Omenn et al., 1980; Passarge et al., 1984; Shashidhar Pai, 1986; Tice et al., 1980), studies in the last 20 years have substantially expanded their focus. Many empirical studies focus not only on this age group, but also on perspectives of parents, siblings and professionals. Some studies investigate genetic counselling and testing (e.g. impact of genetic testing, attitudes towards testing, etc.), while others focus on developing protocols, recommendations and/or ethical principles appropriate for working with young people.

Many young people seem to be in need of additional information and guidance from professionals (e.g. online resources where they can read more about genetic conditions, access to

support groups, access to the genetic counsellor for additional information etc.) (Housten et al., 2015; Markova et al., 1984; MacLeod et al., 2013; Rutherford et al., 2014; Szybowska et al., 2007). Parents' needs mainly include guidance on how to tackle strong emotions (e.g. guilt, blame, shame etc.); how to manage family relationships and how to communicate sensitive information (e.g. genetic conditions, carrier status or family history) (Andrighetti et al., 2015; DeMarco et al., 2008; Haakonsen Smith et al., 2017; Hill et al., 2018; Maloney et al., 2012; Tercyak et al., 2007). Genetic counsellors perceive children and adolescents as a distinct patient group and are challenged to develop new strategies to work with children and adolescents; they point out the need for additional training and versatile and adaptable protocols (Duncan & Young, 2013; Tse et al., 2013; Ulph et al., 2010).

The main conclusions of several systematic reviews (Borry et al., 2005; Borry et al., 2007; Godino et al., 2015; McGill et al., 2018; Rew et al., 2009; Vetsch et al., 2018; Wakefield et al., 2016) are that the literature on this topic is under-researched, divided and heterogeneous in terms of methodologies and recommended that evidence-based guidelines for future practice are needed (Borry et al., 2007; Vetsch et al., 2018; Wakefield et al., 2016).

## 3. Genetic counselling for children and adolescents with psychiatric disorders

A relatively new area of research and practice in genetic counselling is psychiatric genetic counselling. Latest estimates indicate that more than 1 billion people worldwide suffer from some form of mental health problems (The Lancet Global Health, 2020). For children and adolescents, approximately 14% of 10 to 19 years olds suffer from mental disorders (World

Health Organization, 2021). Recent advances in genetic research have provided genetic counsellors with important information which can be used in psychiatric genetic counselling.

### *3.1. Genetics and psychiatric disorders*

Mental health problems have often been regarded as "running in families" (Pestka, 2005). Twin, adoption and family studies have shown that mental health problems have both a genetic and an environmental component (Hill & Sahhar, 2006; Pestka, 2005). Several DNA variations increase the risk for mental health problems (Smoller et al., 2018; Smoller, 2019). These DNA variations include single nucleotide polymorphisms (SNPs), rare copy-number variants (CNVs), mutations in specific genes (e.g. CHD8, SCN2A, SHANK3, GRIN2B for autism spectrum disorders) and rare, de novo variations (Smoller et al., 2018). Psychiatric conditions are also highly polygenic and genetic overlap between disorders has been identified (Smoller et al., 2018; Smoller, 2019). So far, several environmental risk factors have been identified: prenatal factors (e.g. premature birth, complications at birth), pregnancy factors (e.g. several infectious diseases, exposure to heavy metals and toxins, preeclampsia etc.), post-birth factors (e.g. low birth weight), life factors (e.g. drug consumption, smoking, vitamin D deficiency, adverse life events, abuse etc.) (Arseneault, 2017; Davis et al., 2016; Rai et al., 2012; Uher & Zwicker, 2017; Vassos et al., 2016).

The American National Society of Genetic Counsellors recommends that psychiatric genetic counselling should be offered for affective disorders (e.g. depression, bipolar disorder), anxiety disorders, obsessive compulsive disorder, schizophrenia, schizoaffective disorder, Alzheimer disease, ADHD, autism spectrum disorders, alcohol dependence (Inglis et al., 2015; Austin, 2019).

### *3.2.Genetic counselling for psychiatric disorders*

A recent meta-analysis (Moldovan et al., 2017) investigating the efficacy of psychiatric genetic counselling showed it has a moderate effect size, which is also maintained at follow-up. Psychiatric genetic counselling is effective in improving knowledge (e.g. information about aetiology, causes, etc.) and psychological outcomes (e.g. emotional distress, guilt, empowerment) (Moldovan et al., 2017). Similar result were obtained by other studies which concluded psychiatric genetic counselling significantly improves empowerment (Gerrard et al., 2020; Inglis et al., 2015; Semaka & Austin 2019), self-efficacy (Inglis et al., 2015), risk perception accuracy (Hippman et al. 2016), reduces symptoms of psychosis (with no change to treatment adherence) (Morris et al., 2021) and feelings of guilt and stigma (Semaka & Austin 2019), and can influence the overall perception of control (Hippman et al. 2016). Psychiatric genetic counselling takes a more holistic approach by integrating known genetic and environmental risk factors, as well as other factors that could contribute to the manifestation of psychiatric disorders (Austin, 2019).

### *3.3. Psychiatric genetic counselling for children and adolescents*

The benefits of psychiatric genetic counselling are increasingly being investigated, but to date, the empirical data available is solely reflecting its impact on adult patients (Inglis et al., 2015; Gerrard et al., 2020; Hippman et al. 2016; Moldovan et al., 2017; Morris et al., 2021; Semaka & Austin 2019). Clearly, psychiatric genetic counselling is very likely to have benefits for younger patients, especially considering the rise in prevalence of mental health problems amongst children and adolescents and the early onset of many psychiatric disorders (e.g. depression, bipolar disorder, anxiety, obsessive-compulsive disorder). This service has the potential to help

young patients and their parents to access multidisciplinary and personalised care (Gonzalez et al., 2015; Ryan et al., 2015), which can help them better understand and adapt to the causes and implications of their condition (Andrighetti et al., 2015; Haakonsen Smith et al., 2017; Hens et al., 2016), yet no empirical studies are currently available.

## 4. Objectives of research

Genetic counselling is a well-established and empirically supported service, with precise clinical guidelines, protocols and ethical frameworks. Genetic counselling is traditionally offered to adult patients. Children and adolescents are increasingly starting to access this service and the benefits for this age group are currently being documented. Several empirical studies, guidelines and recommendations have been published, but the literature is scarce and heterogenous.

The main aim of this thesis is to assess the impact of genetic counselling for children and adolescents, with particular focus on psychiatric disorders and psychiatric genetic counselling. In order to achieve this aim, the specific goals of the thesis are:

1) To systematically review the scientific literature addressing genetic counselling for children and adolescents.
2) To explore in depth the perspectives (e.g. perceptions, experiences, needs) of careers and young people with complex needs and professionals working with them.
3) To investigate the impact psychiatric disorders have on caregivers (e.g. burden, stigma, well-being).
4) To explore the current practices and needs of genetic counsellors and other healthcare professionals working in genetic counselling with children and adolescents.

5) To investigate the efficacy of psychiatric genetic counselling for parents of children and adolescents diagnosed with psychiatric disorders.

## Chapter II. Methodology

This chapter presents in detail the methodology used for each of the five studies conducted for this thesis. In Study 1 we performed a systematic review where we assessed the current state of the literature. Study 2 focused on investigating the perceptions, needs and experiences of the main stakeholders impacted by complex health needs. In Study 3 we explored the impact psychiatric disorders have on caregivers and we identified the main predictors of burden, stigma and well-being. Study 4 was aimed at exploring the current international practices in genetic counselling for children and adolescents. Study 5 incorporated the results and conclusions of the previous studies: we adapted and implemented an online protocol of psychiatric genetic counselling for parents who have children and adolescents diagnosed with a psychiatric disorder.

### 1. Systematic review

The rapid advances in science can be seen in the vast number of research published every day. Systematic reviews are "a methodical, comprehensive, replicable, and transparent approach" (Siddaway et al., 2019, p.9.5), aimed at identifying, examining and integrating specific evidence, which in turn can inform and guide theory, practice and policy makers (Muka et al., 2019; Siddaway et al., 2019). This type of methodology can allow for a clear synthesis and a critical analysis of the quality of research on a particular topic, construct or research question, by also providing an overview of the current state of the research and of current gaps which need further investigation (Siddaway et al., 2019).

To our knowledge, no systematic review or meta-analysis on genetic counselling of children and adolescents has assessed the current state of the literature so far. We decided to

conduct a systematic review in order to identify, appraise and summarise the existing scientific evidence on genetic counselling of children and adolescents published until 2019, as well as to outline potential research gaps which may need to be addressed in future studies [Study 1].

**2. Thematic analysis**

Qualitative research is key for the "the exploration of meanings of social phenomena as experienced by individuals themselves, in their natural context" (Malterud, 2001, p. 483). Thematic analysis is one of the most commonly used methods in exploring people's perceptions, needs, expectations, experiences and representations. It "identifies, analyses and reports patterns (themes) within data" (Braun & Clarke, 2006, p.79). Themes allow for an accurate description of a complete data set or to focus specifically on a particular aspect of interest related to a specific research question (Braun & Clarke, 2006).

For this thesis, a qualitative study was conducted with the purpose of exploring carers' and young people's experiences and needs when living with long term conditions, as well as the views of the healthcare professionals supporting them. By means of data source triangulation (Carter et al., 2014), 30 semi-structured interviews were conducted with 11 healthcare professionals, 10 parents and 9 young people. Purposive sampling was used to recruit the participants and was based on their experience working with families with complex needs (i.e. health professionals) or on their diagnoses or complex needs or their caregiver role (i.e. parents and young people). Following the interviews, the data gathered was analysed using thematic analysis [Study 2].

### 3. Regression analysis and prediction models

Multiple linear regression analysis is a versatile and valuable research method, through which different associations between a continuous outcome and multiple predictors can be investigated (Stoltzfus, 2011). By carefully selecting relevant and statistically significant predictors and investigating their relationship with the outcome variable, integrative predictive models can be created. Although this is a statistical strategy, results from predictive models can help guide, create and implement research and clinical practice towards a deeper and better application of evidence-based science (Kattan & Gerds, 2020; Rocca & Yarkoni, 2021; Stoltzfus, 2011).

In the current research, in order to better understand how caregivers are impacted by their family members' diagnosis of a psychiatric disorder, we investigated the predictors of caregivers' experienced burden, stigma and well-being. The statistically significant predictors were included in predictive models which can help create targeted interventions specifically for family members [Study 3].

### 4. Survey

Among the large variety of research methodologies available, one particularly popular method is the survey. Most often, this method is used when researchers want to gather information from a large sample of individuals, based on their answers to specific questions (Ponto, 2015). Two important principles are: (1) recruitment of a random, but representative sample of the population of interest, based on specific characteristics (e.g. age, gender, educational background, socioeconomic traits, profession etc.) which can accurately reflect and allow for application of the results to the entire population; (2) selection of appropriate data

collection methods (i.e. instruments used, administration method), which are in accordance with the research question and which help prevent measurement errors (e.g. error of reliability or validity) (Ponto, 2015).

For the current research, an exploratory survey was conducted. Genetic counsellors who have worked with children and adolescents from several countries were invited to participate. The purpose of the study was to investigate current practices, knowledge, skills and self-efficacy in the counselling process of children and adolescents and current training status and needs of genetic counsellors [Study 4].

**5. Pilot intervention**

Ideally, before conducting a randomised control trial, a pilot study is needed in order to investigate several aspects about the intervention investigated (Bell et al., 2018; Thabane et al., 2010). A non-randomized pilot study can help assess if the implementation of a certain intervention (e.g. psychiatric genetic counselling of parents) can be best done. This type of study can help inform on all other aspects of a future randomised trial: recruitment of targeted population, potential randomization, retention of participants, adherence to intervention, fidelity of intervention, investigation of methods of assessment (e.g. time needed, instruments used etc.), acceptability of intervention, costs and benefits of intervention etc. (Pilot Studies: Common Uses and Misuses, 2015).

For this thesis, a non-randomized pilot study was conducted with the aim of assessing an online psychiatric genetic counselling intervention for parents of young people diagnosed with a mental health problem (e.g. protocol, impact, benefits and costs, instruments used etc.) [Study 5].

# Chapter III. Original research

## 1. Genetic counselling for children and adolescents: A mixed-method systematic review

### Introduction

Genetic counselling is a well-established process and good practice guidelines are available. A growing number of studies indicate that young people increasingly participate in genetic counselling sessions (see Callard et al., 2011; Duncan & Young, 2013; Lynch et al., 2010; Pichini et al., 2016). Before 1990, less than a dozen studies were available, but in the last decade the research has doubled in size.

Recent studies have started to focus on young people and parents' opinions and experiences after genetic counselling (MacLeod et al., 2013; Pichini et al., 2016; Duncan & Young, 2013; Everett et al., 2016; Gaff et al., 2006; Smith et al., 2015). Studies also explore genetic counsellors' challenges, knowledge and confidence when working with children and adolescents (Callard et al., 2011; Gonzalez et al., 2015; Werner-Lin et al., 2018).

The aim of our study is to explore how this area of research has been addressed over time and explore the available evidence documenting the main research outputs and clinical recommendations in genetic counselling for children and adolescents.

### Method

An extensive electronic search was conducted investigating the literature published until January 2019, without a specific starting point, in two databases - PubMed and Cochrane. Search terms used to identify relevant articles were: a) genetic counselling; b) genetic counsellor; c) children;

d) adolescents; e) minors; f) young people; g) parents. Inclusion criteria were studies published in English that: (1) investigate genetic counselling for children and adolescents; (2) explore young peoples' and/or their families' experience with genetic counselling (e.g. siblings, parents); (3) address genetic counsellors' opinions and experience of working with children and adolescents; (4) include qualitative or quantitative evidence; (5) discuss recommendations and guidelines regarding the practice of genetic counselling for children and adolescents. Studies were coded to identify: type of article (empirical study, guideline, etc.), research topic (e.g. genetic counselling, genetic testing, communication of risk, etc.), type of participants (e.g. children, adolescents, parents, professionals), number of participants, age of participants, type of genetic disorder (e.g. cystic fibrosis, sickle cell disease, pediatric cancer, etc.), type of evidence (e.g. quantitative, qualitative, mixt), type of analysis (e.g. questionnaire, record screening, thematic analysis etc.) and timeline measurements (e.g. pre-, post- and follow-up).

**Results**

A total of 10.052 studies were identified through database and additional records searching. 147 articles met the inclusion criteria. Figure 1 shows the flowchart of the study selection process.

We grouped the 147 articles in three main categories: articles focusing mainly on (1) young people (49 articles); (2) parents (66 articles); and (3) professionals (69 articles). Each category was then sub-categorized according to type of evidence (quantitative or qualitative) (see Lizarondo et al., 2019; Noyes et al., 2019).

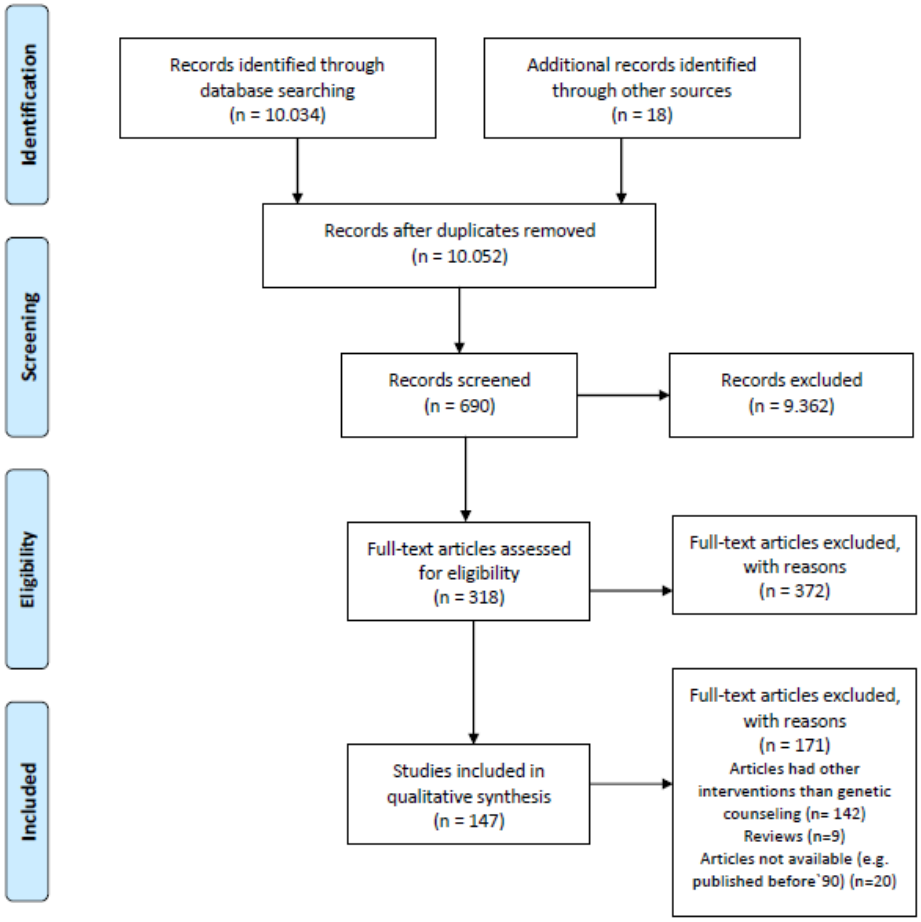


**Figure 1.** Flow diagram of research procedure

## 1. Young people

Of the total of 49 articles, 21 articles focused on young patients, two on young siblings and 26 had mixt participants (e.g. children, adolescents, siblings, parents and/or professionals). Several articles used mixt methods designs, 18 articles used quantitative methods, 17 qualitative methods. Scientific research focusing on counselling children and adolescents has some common areas of interest and are supportive in evidences presented. Studies are investigating attitudes and consequences of genetic counselling and testing in childhood, best practices for genetic

counselling, diagnosis and treatment of young people, best communication strategies with professionals and family members.

Although there are studies investigating genetic counselling, or the impact of multidisciplinary teams of health professionals in genetic counselling, no studies have compared genetic counselling with other types of interventions (e.g. educational sessions, genetic counselling of children/adolescents versus family).

**2. Parents**

From the total of 147 articles included, 66 had as population of interest parents. Of them, 41 focused only on parents and 25 had mixt population (e.g. children, adolescents, siblings, parents and/or professionals). Quantitative methods were used in 38 articles and qualitative in 20 articles. Common topics investigated are parental needs, access, experiences and challenges from genetic counselling/testing, best practices for counselling the family, psychosocial implications of genetic testing for the family and family communication about genetic risk.

From the studies included in this section, we were able to conclude that parents feel it is difficult to balance emotions, disclosure and family relationships, to tolerate the uncertainty of the prognosis and balance family needs, become advocates for their children in accessing support and health services and also deciding what is in the best interest of the child (Duncan & Young, 2013; Hill et al., 2018; Kasparian et al., 2018; MacDonald, & Lessick, 2000; Patenaude & Schneider, 2017).

**3. Professionals**

From 147 articles, 69 articles investigating genetic counselling for children and adolescents were focused mainly on professionals. Of them, 11 had as population professionals and 15 had mixt population (e.g. children, adolescents, siblings, parents and/or professionals). 51 studies focused

on recommendations, guidelines or ethical aspects. Quantitative methods were used in 9 articles and qualitative in 8 articles. Common aspects between studies were related to specialists' practices and strategies in working with under age young people, experiences and challenges in genetic counselling and testing of minors, communication in genetic counselling, management of confidentiality and autonomy of these patients. Studies in this category are more divided than in the previous 2 categories when it comes to methodology, research design, measurements, outcomes and information reported in the article (e.g. age of professionals, years of experience, clinical setting etc.).

**Discussion and conclusions**

Studies investigating genetic counselling for children and adolescents are overwhelmingly **heterogeneous**, not only in terms of population and method of investigation, but also in terms of topics addressed. The majority of evidence available comes from studies investigating parents and/or professionals, and very few from young people.

**Young people** want to understand the science behind their condition, how their condition affects them, and how to manage it at a much younger age than when they usually receive genetic counselling (Eisler et al., 2016; Skotko et al., 2011; Szybowska et al., 2007). **Adolescents and parents** both feel that genetic counselling and testing should be offered before adulthood (Liljestrom et al., 2007) as genetic information and education can help adolescents better adapt to their condition, make informed decisions and foster autonomy (Porter et al., 2014). **Genetic counsellors** perceive children and adolescents as a distinct patient group, who may not be receptive to traditional genetic counselling techniques and who do not understand medical terminology (Duncan & Young, 2013; Tse el al., 2013).

The data we have gathered has provided us with a rich perspective on the main **difficulties and concerns** encountered in genetic counselling of young people. Genetic counsellors as well as young people presented challenges during the counselling process (e.g. creating a therapeutic relationship, communicating during the session, preserving confidentiality and autonomy, balancing family involvement), while parents faced difficulties that arise not only in the counselling process but also before and after the session itself.

Genetic counselling of young people is complex and it involves multiple aspects, from ethical to practical, from individual to familial, from specific practices to general guidelines. The large numbers of studies identified in our research show a great interest in this topic as well as a need for future targeted studies, to help us better understand each aspect of this diverse and multifaceted process.

## 2. Families with complex needs: an inside perspective from young people, their carers, and healthcare providers[1]

### Introduction

Complex needs most often involve additional medical, psychological or social support; long term personalised care; constant re-evaluation, adaptation and management of care planning; involvement of several and often new stakeholders; accessing appropriate health, educational and community services; adapting family life according to each member's needs (Brenner et al., 2018; Brenner et al., 2021; McGregor et al., 2016). All stakeholders involved (e.g. patients, carers, healthcare professionals) face a unique set of challenges and needs. Parents have to balance family dynamics, overcome financial difficulties, advocate for adequate health care (Hill et al., 2018; Kasparian et al., 2018; McGrath et al., 2009; Patenaude & Schneider, 2017; Smith, Oswald & Bodurtha, 2015; Woodgate et al., 2015). Young people's require constant transitions, transformations and re-adaptations of care (Brenner et al., 2021; Kirk, 2008). Healthcare professionals are constantly searching for better ways to support families with complex needs (Bradford et al., 2018; Mattson & Kuo, 2018) by developing or adapting services to appropriately meet families' needs (Duncan & Young, 2013; Gaff et al., 2006; Herman & Appelbaum, 2010).

The aim of the study was to explore carers and young people's needs in the context of living with long term conditions as well as the views of the healthcare professionals supporting them.

[1] Published article: Radu, M., Moldovan, R., & Băban, A. (2022b). Families with complex needs: an inside perspective from young people, their carers, and healthcare providers. Journal of Community Genetics. https://doi.org/10.1007/s12687-022-00586-z

## Method

We conducted 30 semi-structured interviews using a data source triangulation method (Carter et al., 2014) to collect a comprehensive set of diverse experiences and to ensure data saturation. We interviewed 11 healthcare professionals, 10 parents and 9 young people. Participants` characteristics are summarised in Table 1. The interview guide was focused on 3 main areas: (1) medical needs (e.g. knowledge about diagnosis, previous experience with healthcare, type of services accessed); (2) psychological needs (e.g. coping strategies and social support); and (3) communication needs (e.g. communication with stakeholders; family dynamics and relationships). In-depth interviews were conducted individually, face-to-face or over the phone. All participants signed an informed consent form. On average, the interviews had a duration of approximately 30 minutes. Interviews were audio-recorded and transcribed verbatim. Thematic analysis was used to identify the main themes in the interviews (Braun & Clarke, 2006; Carter et al., 2014).

**Table 1:** Participants' characteristics

| Participant | Age | Gender | Young people's diagnosis | Profession, years of experience (for Professionals) |
|---|---|---|---|---|
| HP1 (Health Professional) | 37 | F | NA* | Pediatric neurologist, 6 years |
| HP2 | 25 | F | NA | Geneticist, 3 years |
| HP3 | 30 | F | NA | Genetic counsellor, 8 years |
| HP4 | 45 | F | NA | Child Psychologist, 5 years |
| HP5 | 29 | F | NA | Child Psychologist, 5 years |
| HP6 | 28 | F | NA | Genetic counsellor, 4 years |
| HP7 | 47 | M | NA | Geneticist, 28 years |

| | | | | |
|---|---|---|---|---|
| HP8 | 39 | F | NA | Genetic counsellor, 8 years |
| HP9 | 32 | F | NA | Child psychiatrist, 6 years |
| HP10 | 45 | F | NA | Nurse, 22 years |
| HP11 | 26 | F | NA | Psychologist from pediatric oncology, 4 years |
| P1 (Parent) | 38 | F | Intellectual disability | Personal assistant |
| P2 | 46 | F | Intellectual disability & epilepsy | Personal assistant |
| P3 | 38 | F | Intellectual disability & ADHD** | Administrative assistant |
| P4 | 38 | F | Autism Spectrum Disorder | Unemployed |
| P5 | 43 | F | Jacobsen syndrome | Personal assistant |
| P6 | 43 | F | Down syndrome | Personal assistant |
| P7 | 55 | F | Prader-Willi syndrome | NGO director |
| P8 | 51 | M | Fragile X syndrome | Store manager |
| P9 | 41 | F | Phenylketonuria | NGO manager |
| P10 | 28 | F | Hydrocephaly, cerebral paralysis & epilepsy | Maternity leave |
| YP1 (Young person) | 35 | F | Spinocerebellar Ataxia | Unemployed |
| YP2 | 28 | B | Hemophilia | Volunteer |
| YP3 | 19 | F | Jacobsen syndrome | Unemployed |
| YP4 | 28 | F | Intellectual disability | Unemployed |
| YP5 | 26 | F | Intellectual disability & epilepsy | Unemployed |
| YP6 | 26 | F | Down syndrome | Unemployed |
| YP7 | 33 | F | Prader-Willi syndrome | Unemployed |
| YP8 | 34 | F | Epilepsy & motor dysfunction | Volunteer |
| YP9 | 28 | B | Achondroplasia | Volunteer |

*NA: Not applicable; **ADHD-Attention deficit hyperactivity disorder

## Results

Following the thematic analysis, four main themes emerged: (1) *Acceptance takes time*; (2) *Close guidance*; (3) *Open communication*; and (4) *Long-term support*.

### 1. Acceptance takes time

Healthcare professionals believe the moment of diagnosis can be overwhelming. All professionals interviewed witnessed parents experiencing intense emotions (e.g. frustration, anger, guilt, helplessness and grief). Stigma was often mentioned as interfering with and prolonging the acceptance and adaptation process, for both parents and young people.

Many parents said they needed time to come to terms with the condition. Most said that, at the moment of diagnosis, they felt anger, sadness, pain, uncertainty, and fear of the future. Some experienced shame and felt excluded from various social or educational activities. Adaptation became easier after having contact with families in the same situation and after learning more about the condition, and how to manage it.

Young people mentioned needing time to accept their diagnosis and the implications that come with it. Almost all those interviewed said they experienced prejudice at some point in their life. Acceptance became easier after meeting other people with similar conditions or needs (people who have an “inside perspective”).

### 2. Close guidance

Professionals mentioned hearing parents say they could have used better guidance in understanding and adapting their expectations during the long and challenging process of finding

the best care for their child. Professionals believed that the main reason carers don't always access the appropriate services is because they “get lost along the way”. Genetic counselling would facilitate a better understanding of and adaptation to the diagnosis and it would also help manage negative emotions.

Most parents had to wait a long time until a final diagnosis. Careers mentioned having difficulties in understanding the process and navigating the health system; that the ideal solution would be a one stop shop, a one stop service that could oversee their journey and guide them throughout. Parents also perceived genetic counselling could have helped them understand the diagnosis, it could have facilitated decisions about family planning and informed healthcare strategies.

Young adults said they could have used genetic counselling when they were younger and started to have questions about their diagnosis. They believed it would have benefited both them and their parents in terms of information, guidance and long term support.

**3. Open Communication**

Carers seemed to perceive professionals as the main gatekeepers of information about diagnosis, access to medical services and long term care. When discussing communication as a family, parents see it very much as a long term process, meaning that they tend to talk to their children about their condition gradually, in time.

Several young people mentioned learning about their diagnosis when they were little, from their parents. Others said they did not talk to their parents about it, they rather learned about it in time, on their own, from books or from the internet.

Professionals said they mostly discussed the condition with the parents; they seemed to perceive carers as the main gatekeepers of information in the family, especially when the affected child is young.

**4. Long-term support**

Almost all parents and affected youth said that the greatest support comes from other family members (e.g. partners, grandparents, aunts, uncles, siblings). They relied on the extended family for emotional, social, practical and financial support.

Young people also mentioned that having childhood friends and developing close friendships with other young people who have the same diagnosis as them, as another meaningful source of support.

Professionals believe families need constant and personalised support. They feel support groups, for instance, are a source of information on non-medical aspects, like education, career options, sexual education, romantic relationships etc.

**Discussion**

Our study was aimed at exploring parents' and affected young people's needs in the context of living with long term conditions as well as the views of the healthcare professionals supporting them. Following the thematic analysis, four main themes emerged: (1) *Acceptance takes time* refers to the often long and challenging process of adapting to the diagnosis and implications of a condition; (2) *Close guidance* captures the importance of specialised and long term support in understanding and accessing the medical system and healthcare services available; (3) *Open communication* shows the families' needs to be closely connected with healthcare providers and

other families dealing with similar difficulties; and (4) *Long-term support* underlines the importance of long term formal and informal support strategies.

By concatenating the views of affected young people, their parents and healthcare providers in the context of complex conditions, we have obtained an in-depth understanding of their experiences and needs. Following a diagnosis that is often unexpected, unpredictable or overwhelming, both families and professionals feel that close guidance, long-term support and open communication contribute significantly to their adaptation and well-being. Genetic counselling, explicitly mentioned by some professionals or tentatively described by some parents or young adults, is an example of a service that can address these needs and contribute with the appropriate support for families living with complex conditions. These findings can provide guidance for developing and implementing more personalised and integrated services (genetic counselling being one example), as well as support future research aimed at better understanding the impact of complex conditions and meaningful ways to support families throughout their journeys.

## 3. The impact of psychiatric disorders on caregivers: An integrative predictive model of burden, stigma and well-being[2]

### Introduction

Looking after a family member diagnosed with a psychiatric condition, especially for those who become main caregivers, is a complex task as it involves providing dedicated and often demanding or challenging assistance and support. (Clibbens et al., 2019; Dwyeret al., 1994; Gérain & Zech, 2019). Regardless of the age of the patient (e.g., children, adolescents or adults), the caregiver role is associated with feelings of burden, health-related difficulties and a lower well-being (Baker et al., 2011; Bom et al., 2018; Estes et al., 2009).

The literature has predominantly focused on the impact chronic conditions (e.g., Down syndrome, autism, sickle cell disease, dementia,) can have on caregivers, especially in terms of burden, quality of life and well-being (Adelman et al., 2014; Barros et al., 2017; Carden et al., 2016; Jeyagurunathan et al., 2017; Macedo et al., 2015; Sheng et al., 2018). Recently, other variables such as illness perception or stigma about mental health have become topical (Han et al., 2021; Hinshaw, 2005; Zhang et al., 2018).

We believe this study can help tailor future psychosocial interventions for family members of patients with psychiatric conditions. Psychiatric genetic cunselling is undoubtedly a promising option for families as it specifically targets understanding and adaptation to conditions with familial implications (Resta et al., 2006).

[2] Published article: Radu, M., Ciucă, A., Crișan, C., Pintea, S., Predescu, E., Șipos, R., Moldovan, R., & Băban, A. (2022a). The impact of psychiatric disorders on caregivers: An integrative predictive model of burden, stigma, and well-being. Perspectives in Psychiatric Care. https://doi.org/10.1111/ppc.13071

We aimed to investigate the main predictors of caregiver's experienced burden, stigma and well-being when looking after family members diagnosed with a psychiatric disorder.

## Method

Participants had to be the main caregivers (e.g., spouses, parents, grandparents, legal guardians) of an adult or young person diagnosed with a psychiatric condition. Family members were diagnosed (as per DSM-V) with schizophrenia, bipolar disorder, schizoaffective disorder, depressive disorder, alcohol use disorder or neurodevelopmental disorders (autism spectrum disorders, Attention-Deficit and Hyperactivity Disorder, intellectual disabilities). Measures included were: (1) The demographic questionnaire; (2) Involvement Evaluation Questionnaire (IEQ, van Wijngaarden et al., 2000); (3) Self-Stigma in Relatives of People with Mental Illness Scale (SSRMI, Morris et al., 2018); (4) The Psychological General Well-Being Index (PGWBI, Dupuy, 1990; Grossi & Compare, 2014); (5) Knowledge (Costain et al., 2014a); and (6) Illness Perception Questionnaire: Relatives' Version (IPQS-Relatives, Lobban et al., 2005). Data collected in this cross-sectional study was analysed to explore the main predictors of caregivers' experienced burden, stigma and well-being.

## Results

### *Demographics*

A total of 168 participants took part in this research, 98 of which were caregivers of adult patients and 70 of which were caregivers of children and adolescents with psychiatric disorders. Details about the participants' demographics are included in Table 1.

**Table 1.** Demographics

| Variable | | n (%) | *m (SD) |
|---|---|---|---|
| **Gender** | Female | 132 (78.6) | |
| | Male | 36 (21.4) | |
| **Ethnicity** | Romanian | 147 (87.5) | |
| | Hungarian | 16 (9.5) | |
| | Other | 5 (3) | |
| **Residence** | Urban | 119 (70.83) | |
| | Rural | 49 (29.17) | |
| **Education** | Less than high school | 51 (30.35) | |
| | Highschool | 64 (38.09) | |
| | Higher education | 53 (31.54) | |
| **Marital status** | Married/serious relationship | 120 (71.3) | |
| | Widow | 13 (7.8) | |
| | Divorced/separated | 17 (10.2) | |
| | Never married | 7 (4.2) | |
| | Single | 11 (6.5) | |
| **Occupational status** | Student | 9 (5.4) | |
| | Unemployed | 30 (17.8) | |
| | Employed | 100 (59.5) | |
| | Retired | 29 (17.3) | |
| **Relationship to patient** | Parent | 75 (44.6) | |
| | Sibling | 18 (10.7) | |
| | Partner | 31(18.5) | |
| | Child | 23 (13.7) | |
| | Cousin | 3 (1.8) | |
| | In law | 5 (3.0) | |
| | Grandparent | 5 (3.0) | |
| | Other | 8 (4.8) | |
| **Diagnostic of patient** | *Adult patients* | | |
| | Schizophrenia | 27 (16.1) | |
| | Bipolar disorder | 7 (4.2) | |
| | Schizoaffective disorder | 1 (0.6) | |
| | Depression | 31 (18.5) | |
| | Alcohol addiction | 32 (19.0) | |
| | *Pediatric patients* | | |
| | Autism spectrum disorders | 20 (12.0) | |
| | ADHD | 26 (15.6) | |
| | Depression | 5 (3.0) | |
| | Bipolar disorder | 2 (1.2) | |
| | Anxiety disorders | 6 (3.6) | |

| | | | |
|---|---|---|---|
| | Psychotic disorders | 3 (1.8) | |
| | Intellectual disabilities | 8 (4.8) | |
| **Age** | | | 47.71 (13.18) |
| **Number of children** | | | 1.65 (1.01) |

N= 168 **m= means, SD= standard deviation*

***Correlation analyses***

*Socio-demographic correlates of burden, stigma and well-being*

Age was positively associated with burden (r= .193, p< .05) and gender was positively associated with both burden (r= .206, p< .05) and stigma (r= .199, p< .05).

Having a parent with a psychiatric condition is positively associated with burden (r= .188, p< .05), whilst being the partner of someone with a psychiatric diagnosis is positively associated with stigma (r= .273, p< .01) and negatively associated with well-being (r= -.159, p< .05). Being a parent of a patient with a psychiatric condition is negatively associated with stigma (r= -.167, p< .05).

*Medical correlates of burden, stigma and well-being*

Our data show that a diagnosis of alcohol use disorder is positively associated with caregivers' burden (r= .167, p< .05) and stigma (r= .227, p< .01).

*Cognitive correlates of burden, stigma and well-being*

The statistical analysis showed knowledge was negatively correlated with stigma (r= -.258, p< .01) and positively correlated with well-being (r= .166, p< .05). For illness perception, correlations were computed for each component of the IPQ scale. The consequences for the

relatives subscale correlates positively with the burden (r= .447, p< .01) and stigma (r= .538, p< .01) and negatively with well-being (r= -.306, p< .01). The emotional representation subscale is positively correlated with both burden (r= .430, p< .01) and stigma (r= .597, p< .01) and negatively with well-being (r= -.383, p< .01).

***An integrative predictive model of burden***

When simultaneously controlling for all relevant factors, the only significant predictors of burden remain gender (β=0.18, p= 0.015) and the emotional representation of illness (β=0.38, p< 0.001), indicating that a higher negative representation of the condition predicts a higher level of burden, particularly in women.

***An integrative predictive model of stigma***

When simultaneously controlling for all relevant factors, the only significant predictor of stigma remains the emotional representation of illness (β=0.53, p< 0.001), which shows that a higher negative representation of the condition predicts a higher level of stigma.

***An integrative predictive model of well-being***

Our results show that when introducing the most relevant predictors of well-being into an integrative model, the only significant predictor remains the emotional representation of illness (β= -0.36, p< 0.001). This indicates that a less negative representation of the condition predicts a higher level of well-being.

## Discussion

Our main aim was to identify the main predictors of caregivers' experienced burden, stigma and well-being when looking after family members diagnosed with a psychiatric disorder.

As expected, burden was associated with stigma, and well-being was negatively correlated with both burden and stigma. We explored in detail the main correlates and predictors of burden, stigma and well-being. The statistical analysis showed caregivers' knowledge about their family member's psychiatric condition impacts the level of stigma and well-being experienced, but not the burden. Additionally, the perceived consequences for the relative and the emotional representation of the relatives' condition were both found to be positively associated with burden and stigma and negatively associated with well-being. Our findings contribute to existing empirical data with evidence showing that illness perception can also impact caregivers' burden and stigma.

The caregivers' emotional representation of the patients' psychiatric condition is the strongest predictor of their burden, stigma and well-being. In order to better understand the caregivers' experience, how to best support them, and how to better address the difficulties they encounter, future research needs to investigate the impact illness perception has on these variables. Psychiatric genetic counselling is a valuable and effective psychosocial intervention that may help tackle and manage these issues, with benefits for both patients and family members.

## 4. Genetic counsellors working with young people: overview of current needs and glimpse forward

### Introduction

Genetic counselling helps improve knowledge about genetic conditions, it contributes to a better management of emotional distress, and facilitates empowerment and patient autonomy (Athens et al., 2017; Moldovan et al., 2017). The overwhelming evidence supporting the efficacy of GC is primarily based on adult data (Athens et al., 2017; Madlensky et al., 2017; Moldovan et al., 2017); recently, a growing number of studies exploring the views of children and adolescents accessing genetic counselling services have been publishd (Callard et al., 2011; Duncan & Young, 2013; Lahlou-Laforêt et al., 2012; Lynch et al., 2010; McGill et al., 2019; Oosterwijk, 2016; Pichini et al., 2016; Stump et al., 2018).

There are several gaps in research when it comes to documenting genetic counselling for young people. We do not have yet an accurate image about current practices or the needs of counsellors. By investigating current practices, we can help tailor genetic counselling training programs and protocols which can better meet the needs of genetic counsellors. The objective of our study is to assess genetic counsellors' knowledge, skills and self-efficacy when delivering the counselling process to children and adolescents; our secondary aim was to explore their current training level and needs.

## Methods

Genetic counsellors, clinical geneticists and other health professionals who have worked in genetic counselling with young people under the age of 18 were invited to participate. An online survey was sent to international professional organisations in Europe, Canada, United States of America, Australia and South Africa. The survey was composed of 5 sections, each investigating a specific aspect of working with young people in genetic counselling: (1) demographic information; (2) current practices (Griswold et al., 2011; Zhou et al., 2014); (3) the Genetic Counsellors Self–Efficacy Scale (GCSES) (Caldwell et al., 2018a); (4) training level (Griswold et al., 2011; Zhou et al., 2014;) and (5) Knowledge (Anghelescu et al., 2010; Dumulescu & Matei, 2018). The data was analysed using descriptive statistics and Pearson correlations.

## Results

A total of 168 answers from 27 different countries were gathered from June 2019 to December 2021. Most respondents were from the United States of America followed by Australia, France, United Kingdom, Canada and Sweden (see Table 1).

**Table 1**. Demographics

| **Demographic information** | | **Number (N=168)** |
|---|---|---|
| Country | Argentina | 1 |
| | Australia | 16 |
| | Austria | 1 |
| | Belgium | 5 |

| | | |
|---|---|---|
| | Canada | 12 |
| | Chile | 1 |
| | Denmark | 2 |
| | France | 16 |
| | Greece | 3 |
| | Iceland | 1 |
| | Ireland | 2 |
| | Israel | 2 |
| | Italy | 1 |
| | Netherlands | 3 |
| | Norway | 3 |
| | Oman | 1 |
| | Pakistan | 1 |
| | Portugal | 2 |
| | Romania | 2 |
| | Saudi Arabia | 2 |
| | Singapore | 1 |
| | South Africa | 7 |
| | Spain | 8 |
| | Sweden | 11 |
| | Switzerland | 1 |
| | United Kingdom | 16 |
| | United States of America | 47 |
| Gender | Male | 12 |
| | Female | 156 |
| Work setting | Public Hospital | 108 |
| | Private Practice | 10 |

| | | |
|---|---|---|
| | University | 23 |
| | Public Hospital & University | 11 |
| | Public Hospital & Private practice | 2 |
| | Public Hospital, Private practice & University | 2 |
| | Other | 12 |
| Job title | Genetic counsellor | 158 |
| | Clinical geneticist | 4 |
| | Nurse | 3 |
| | Other | 3 |
| Professional background | Bachelor of science or genetics | 91 |
| | Bachelor of nursing or midwifery | 18 |
| | Bachelor of psychology | 10 |
| | Other Bachelor degree | 12 |
| | Master's degree in genetic counselling | 137 |
| | Master's degree in genetic nursing | 1 |
| | Other Master's degree | 35 |
| | PhD | 24 |
| | Medical Geneticist | 3 |
| | Other Medical specialty | 10 |
| Worked in genetic counselling with children and adolescents? | Yes | 131 |
| | No | 37 |
| Age (range) | | 38.7 ± 11.2 (24 to 68 years) |
| Years of experience (range) | | 9.8 ± 8.6 (0 to 42 years) |

**Current practice**

From the 131 respondents who reported having worked with children and adolescents, most of them had seen all young age groups. The most reported *source of referral* for services was parents (85%), followed by general health professionals (72%). All participants in the survey said parents tend to *accompany* young people to the session, followed by siblings (24%). The most frequent *reasons for attending* the session were a family history of a genetic condition (78%) or a personal history of a genetic condition (76%). The most frequent *conditions* participants had worked with were cancer syndromes or predispositions, followed by cardiac conditions and chromosomal abnormalities. *During the session*, genetic counsellors reported spending most time discussing testing options/results (77%) and discussing inheritance/recurrence risk (71%). When asked which information they believe young people have a *hard time understanding*, 44% mentioned the diagnosis and evolution of the condition, 37% mentioned the psychosocial support. 60% of the respondents agreed that *the most challenging information to discuss* in session is the diagnosis and evolution of the condition, and 41% mentioned psychosocial support. When working with children and adolescents, 66% of respondents reported the most challenging *aspects to tackle* were engaging the patient in the session, followed by discussing long term consequences (60%).

**Self-efficacy**

The highest mean for the 6 Factors of the Self-efficacy scale was for Factor 3 Genetic testing (70.1 ± 7.9), followed by Factor 2 Communication (67.8 ± 8.8) and Factor 1 Complex Skills (63.6 ± 10.0). The lowest mean scores were for Factor 6 Information gathering (27.9 ± 2.6) and Factor 4 Basic Psychosocial Skills (42.1 ± 5.8).

**Training level**

Almost 70% of genetic counselors agreed their training had provided them with sufficient *theoretical knowledge* to work with young people, with only 2% strongly disagreeing. When asked about their *practical skills*, 31% strongly agreed they had been provided with sufficient, and 1% strongly disagreed. When asked if they feel they have *access to additional training,* approximately half of participants agreed (22% strongly agreed and 30% agreed), and 23% disagreed. In terms of *additional training*, 60% of the respondents felt they would benefit from additional training on performing psychosocial assessment (e.g. decision making support and crisis counselling) and 43% would benefit from skills-training (e.g. communication, non-directivity, empathy).

**Knowledge**

Questions about adolescent development (10 to 18 years old) were answered correctly in a higher proportion than those about children up to 10 years old. Mean scores on the knowledge scale for children up to 10 years old was 15.1 (±4.0). For adolescents between 10 to 18 years old, the mean score was 18.4 (±2.5.0).

**Correlation analyses**

Genetic counselling self-efficacy was positively associated with both age ($r = .23$, $p < .01$) and years of professional experience ($r = .21$, $p < .05$). Training level was negatively associated with Factor 6 Information gathering ($r = -.40$, $p < .01$), with Factor 3 Genetic testing ($r = -.39$, $p < .01$) and with knowledge about children up to 10 years old ($r = -.23$, $p < .01$). Additional training needs were negatively associated with Factor 6 Information gathering ($r = -.28$, $p < .01$) and

Factor 3 Genetic testing (r= -.25, p< .01). Having a PhD was negatively associated both with Factor 3 Genetic testing (r= -.20, p< .05) and Factor 6 Information gathering (r= -.24, p< .01).

**Discussion**

An international survey assessing current practice, self-efficacy, knowledge, training level and needs among genetic counsellors working with young people under the age of 18 was conducted. Most participants were genetic counsellors, who worked with young people of all ages, at the request of general health professionals or paediatricians, who had a family or personal history of a genetic condition. Session included a variety of diagnoses and practical challenges. Counsellors reported the highest level of self-efficacy when discussing genetic testing and the lowest for information gathering and applying basic psychosocial skills. Almost half agreed they have access to additional training if needed. Counsellors also correctly answered more knowledge questions about adolescent development than about child development.

Genetic counsellors are trying to manage their own needs and the ones of their patients, by finding the appropriate framework in which they can deliver science driven as well as easy to understand information. However, our survey has shown current practices vary and genetic counsellors have many practical and theoretical challenges to overcome when working with children and adolescents. By incorporating knowledge from developmental psychology and by working on developing specific, flexible and adaptable counselling skills, genetic counsellors can feel better equipped and empowered to work with children and adolescents. These represent key aspects for future training and supervision programs.

## 5. Genetic counselling for caregivers of children and adolescents with psychiatric disorders - a pilot intervention

### Introduction

Psychiatric genetic counselling is the process through which psychological support and complex information about the aetiology and management of mental disorders can be delivered to patients and their families (Austin & Honer, 2008; Costain et al., 2014a; Costain et al., 2014b). Recent advances in genetic research have provided important insight into the etiopathogenetic mechanisms of mental disorders (Cross-Disorder Group of the Psychiatric Genomics, 2019; Smoller et al., 2018; Smoller, 2019; Sullivan & Geschwind, 2019), offering additional information about the complex interaction between genetic and environmental factors involved in the occurrence of psychiatric disorders (Hill & Sahhar, 2006; Pestka, 2005).

Knowledge about mental disorders, emotional distress, empowerment, self-efficacy and recurrence risk perceptions are among the main outcomes improved after attending psychiatric genetic counselling (Gerrard et al., 2020; Inglis et al., 2015; Hippman et al. 2016; Moldovan et al., 2017; Semaka & Austin 2019). Recent studies have already started tailoring psychiatric genetic counselling for parents, children and adolescents (Griesi-Oliveira& Sertié, 2017; Haakonsen Smith et al., 2017; Hoang et al., 2018). However, the impact of these interventions has not yet been investigated. This exploratory study aims at investigating the benefits of psychiatric genetic counselling for parents of children and adolescents diagnosed with a psychiatric disorder.

## Method

Parents of children and adolescents diagnosed with mental disorders were invited to participate. Parents had to: (1) be over the age of 18 and be the primary caregivers of a child/adolescent diagnosed with a psychiatric disorder; (2) the child/adolescent had to be diagnosed with affective disorders, anxiety disorders, obsessive-compulsive disorder, psychotic episodes or neurodevelopmental disorders (e.g. ADHD, autism spectrum disorders, intellectual disabilities); (3) have official documents (e.g. medical letter, medical records, etc.) in which the diagnosis was confirmed. Each parent had to fill in (via Google Forms), pre- and post-intervention, five questionnaires: (1) demographic information; (2) The Genomics Outcome Scale (GOS, Grant et al., 2019); (3) Knowledge (Costain et al., 2014a); (4) Self-Stigma in Relatives of people with Mental Illness Scale- the ten-item version (SSRMI, Morris et al., 2018) and (5) The Psychological General Well-Being Index- the short version (PGWB-S, Grossi et al., 2006). Each participant attended a single online session of psychiatric genetic counselling, which lasted ~1.5 hours and the structure of the session was similar to genetic counselling for other disorders. Data analysis included descriptive statistics, paired sample t-test were, difference scores (i.e. delta change) and Pearson correlation.

## Results

A total of 43 parents voluntarily decided to take part in this study, 36 of which attended the genetic counselling session, and 32 completed the post-counselling questionnaires. Most participants were female (95%), with an average age of 40 years old (±5.9) and were the main caregivers of children and adolescents diagnosed with ASD (67.4%), ADHD (23.3%), affective

disorders (7%) and anxiety disorders (2.3%). Detailed information about participants' demographics are presented in Table 1 and Table 2.

**Table 1.** Demographics

| Variable | | Number (%) |
|---|---|---|
| **Gender** | Male | 2 (4.7) |
| | Female | 41 (95.3) |
| **Marital status** | Married | 38 (88.4) |
| | Widow | 5 (11.6) |
| **Ethnicity** | Romanian | 41 (97.7) |
| | Other | 1 (2.3) |
| **Education** | Less than high school | 1 (2.3) |
| | High school | 10 (23.3) |
| | Higher education | 32 (74.4) |
| **Occupational status** | Unemployed | 7 (16.3) |
| | Employed | 28 (65.1) |
| | Other | 8 (18.6) |
| **Residence** | Urban | 31 (72.1) |
| | Rural | 12 (27.9) |
| **Number of household family members** | Two | 2 (4.7) |
| | Three | 15 (34.9) |
| | Four | 19 (44.2) |
| | Five | 5 (11.6) |
| | Over five | 2 (4.7) |
| **Religion** | Orthodox | 40 (93.0) |
| | Catholic | 1 (2.3) |
| | Not religious | 2 (4.7) |
| **Monthly Income** | Under 2000Lei | 12 (28.0) |
| | 2000-3000Lei | 7 (16.3) |
| | 3000-4000Lei | 9 (20.9) |
| | Over 4000Lei | 15 (34.9) |
| **Main diagnostic of child or adolescent** | ASD* | 29 (67.4) |
| | ADHD** | 10 (23.3) |
| | Affective Disorders | 3 (7.0) |
| | Anxiety Disorders | 1 (2.3) |
| **Comorbidities** | None | 17 (39.5) |
| | ADHD** | 12 (27.9) |
| | Anxiety Disorders | 4 (9.3) |
| | Learning disabilities | 4 (9.3) |
| | Intellectual disabilities | 4 (9.3) |
| | Other | 2 (4.7) |
| **Total family members with psychiatric disorders (besides child/adolescent)** | Zero | 26 (60.5) |
| | One | 11 (25.6) |
| | Two | 4 (9.3) |
| | More than three | 2 (4.7) |

N= 43, *ASD: Autism Spectrum Disorders, **ADHD: attention deficit hyperactivity disorder

*Paired sample t-test*

The results indicate a significant difference between parents' pre-test knowledge (M= 8.38; SD= 4.6) and post-test knowledge (M= 13.13; SD= 4.1); t(31)= -7.132, p=.000, following the session. The same significant difference was seen between pre-test empowerment (M= 20.69; SD=3.1) and post-test empowerment (M=22.28; SD= 2.7); t(31)= -2.723, p= .011.

*Delta change and Pearson correlation*

Pearson correlation was performed between delta change scores for each variable and several demographic variables. Delta change for knowledge was positively associated with anxiety disorders (r= .400, p< .05), suggesting that parents who have children with anxiety disorders benefit from psychiatric genetic counselling in improving their knowledge; delta change in stigma was positively associated with ASD (r= .379, p< .05), indicating that parents who have children with ASD benefit from genetic counselling on improving their experienced stigma. The year of diagnosis was positively associated with delta change for knowledge (r= .388, p< .05) and well-being (r= .381, p< .05), meaning that the sooner after the moment of diagnosis parents access genetic counselling, the greater it impacts their knowledge and well-being.

**Discussion**

The main aim of this pilot study was to investigate the impact of psychiatric genetic counselling on parent's knowledge, empowerment, well-being and stigma. As hypothesised, the statistical analysis revealed that parents' levels of knowledge and empowerment changed after

attending the genetic counselling session. When conducting Pearson correlations between changes in stigma and each diagnosis, results showed that parents of children and adolescents with ASD benefited in terms of lowering their experienced stigma after attending the session. The statistical analysis also revealed other interesting aspects, like for example the least amount of time passed from the moment of diagnosis until attending the counselling session, the more it impacts parents' levels of knowledge and well-being.

This study was to our knowledge the first to implement psychiatric genetic counselling for parents of children and adolescents with psychiatric disorders. Clearly, psychiatric genetic counselling is a valuable process with specific benefits; it helps patients and their families better understand the mechanisms of mental disorders as well as strategies which they can use to protect their own mental health (Austin, 2019; Inglis et al., 2015; Maio et al., 2013; Moldovan et al., 2019).

# Chapter IV. Conclusions

## 1. General conclusions

In this thesis we focused on investigating genetic counselling for young people and their families, by integrating the perspectives and needs from all stakeholders involved (e.g. children, adolescents, parents, health professionals) and by documenting and implementing a counselling intervention building on empirical data and clinical guidance available. The first study was aimed at reviewing the current state of published literature on genetic counselling for children and adolescents. The second and third studies investigated through qualitative and quantitative methods the needs (i.e. medical, informational, communicational and psychosocial) and the emotional distress (e.g. levels of stigma, burden, well-being) experienced by families with complex health needs. The fourth study explored current practices and needs of genetic counsellors working with children and adolescents. The fifth and final study was designed to investigate the impact of genetic counselling for parents of children and adolescents with psychiatric conditions.

This thesis contributes with several important contributions to the field: (1) theoretical, through the systematic review, (2) practical, by better understanding the needs of all stakeholders involved (e.g. young people, parents, genetic counsellors), and (3) clinical, by documenting the efficacy of psychiatric genetic counselling for parents who have children diagnosed with psychiatric conditions.

## 2. Specific key conclusions

### 2.1. Systematic review of genetic counselling of children and adolescents

A systematic review investigating the current state of the literature on genetic counselling for children and adolescents was conducted. Results reveal that although the research on this topic has doubled in size in the last decades, studies are still overwhelmingly heterogeneous in terms of population, methods of investigation and topics addressed. Both quantitative and qualitative studies have concluded that the request and need for genetic counselling for young people with genetic diseases has significantly increased.

This is the first systematic review to incorporate all existing literature on genetic counselling for children and adolescents. Due to the heterogeneity of the literature, our original intention to do a meta-analysis was not achieved; it also proved difficult to organise the studies in distinct categories. Our study could inform future empirical studies focused on various aspects relevant for genetic counselling and contribute to creating a more cohesive scientific area of research, which will help in the long run to develop appropriate counselling protocols and training programs.

### 2.2. Inside view of families with complex needs

This study focused on exploring the views of caretakers and young people living with complex care needs, as well as the perspectives of the healthcare professionals supporting them. Through semi-structured interviews and thematic analysis, four main themes were identified: *acceptance takes time, close guidance*, *open communication* and *long-term support*.

By presenting these views together, we were able to gain a better understanding of the challenges these families face, as well as exploring new-emerging services (e.g. genetic counselling) which can be a significant pillar in their long-term care.

Future investigation should explore in depth the perspectives of other family members impacted by complex health care needs, like for example fathers or siblings, who often become co-careers. Also, studies looking into how to best adapt and implement new-emerging services (e.g. genetic counselling) in specific social-cultural contexts (e.g. Romania) in order to implement state-of-the-art services are needed.

### 2.3. An integrative predictive model of burden, stigma and well-being for caregivers of psychiatric patients

This cross-sectional study assessed socio-demographic and cognitive correlates of burden, stigma and well-being experienced by main caregivers of individuals diagnosed with psychiatric conditions. After conducting a statistical analysis for integrative predictive models for all main variables, results showed that a higher negative representation of the condition predicts a higher level of burden and stigma, while a less negative representation of the condition predicts higher levels of well-being.

Our research complements previous studies with the prediction models conducted as it helped highlights a new predictor (i.e. the emotional representation of the condition), which adds a new important variable to the ones already documented. When extrapolating the results, conclusions should be interpreted with caution due to the relatively small sample size. At the same time, other variables (e.g. quality of life) or other components of illness perception (e.g.

consequences, personal control, etc.) might be relevant factors which could contribute to the overall adaptation to the condition of family members of patients with psychiatric disorders.

### 2.4. Current practices and needs of genetic counsellors working with young people

The aim of this study was to investigate current practices, training status, training needs, self-efficacy and knowledge of genetic counsellors working with young people under the age of 18. To our knowledge, this is the first study who has investigated current practices and needs of genetic counsellors working with young people. We have now collected additional information about the context and structure of these sessions (e.g. who attends, who makes the referral, how the session is structured etc.), and what are genetic counsellors' needs (i.e. theoretical knowledge and practical skills) in order to better provide this service.

One important limitation of this study is that the questions assessing knowledge were developed for the purpose of this study and need further investigation and validation.

*Based on this* exploratory study, more detailed investigations of all the variables in this study are needed. Also, development of standardised assessment tools would add great value to future studies and training programs as they are clearly essential for the development of evidence–based practices.

### 2.5. Psychiatric genetic counselling for parents of young people with psychiatric conditions

The impact of psychiatric genetic counselling on parents' knowledge, empowerment, well-being and stigma was assessed in this pilot study. Results showed that psychiatric genetic counselling impacted parents in several ways: their levels of knowledge and empowerment improved compared to their pre-session scores. More so, the less time passed from the moment of diagnosis until attending the session, the more the session impacted their knowledge and well-being. These findings complement existing empirical data showing that family members of adult patients with psychiatric disorders benefit from psychiatric genetic counselling; it is now clear that these benefits can be seen when offering psychiatric genetic counselling to parents whose children were diagnosed with psychiatric disorders.

However, results should be interpreted with caution due to the low the statistical power and the small sample size. Future research can further validate the results of this pilot study, by investigating parents' benefits after attending psychiatric genetic counselling in randomised control trials.

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