Mentoring Guide

1. Introduction

Year 1984 marked the beginning of an effort to decode the human genome (Human Genome Project); an effort that was completed in 2003 and significantly contributed to a better diagnostic approach of many genetic diseases. The use of industrialised methods of data analysis and correlation improved the diagnostic accuracy and therapeutic approach of these diseases. This meant the transition to molecular karyotype (a-CGH), panels and next-generation sequencing techniques. These techniques accomplish the sequencing of thousands to millions DNA bases and mark a new era in the diagnosis of genetic diseases.

Assessing the personal and family medical history, as well as the phenotype of the patient, Clinical Medical Genetics uses the technology of specialised genetic tests to offer diagnosis, prognosis, prevention, monitoring and genetic counselling for patients.

In the world of medicine and pharmaceutics, the traditional “one-size-fits-all” approach is gradually being replaced by the so-called “precision” or “personalized” medicine. Critical clinical decisions may be based on the genetic, epigenetic, proteomic and metabolomic profile of each patient.

2. Personalized prevention

Recent advances in molecular genetics and genetic engineering are used to reshape the whole pharmacogenetics landscape in terms of managing rare genetic diseases and highlight the importance of highly accurate genetic testing and analysis. The characterization of DNA variants and the identification of the underlying mechanisms foster the development of novel targeted pharmaceutical approaches. Next Generation Sequencing (NGS) technologies allow simultaneous testing for more than

one gene or even the whole exome/genome, providing a precise genomic profile for each patient. The so-far appraisal of NGS technologies in diagnosis of rare genetic diseases allows the disclosure of a constantly increasing number of variants in known or novel genetic loci.

Personalized prevention acknowledges that each genetic profile is shaped by DNA variants and differentiates each patient's risk of disease and disease severity, as well as their likely response to preventive or therapeutic interventions. In general, genetically-based therapies target the genetic components of a cell, namely the DNA or RNA, and include gene therapy with the incorporation of normal genetic regions (the whole gene or part of it), genome editing where DNA modifications take place, RNA modifications (AntiSense Oligonucleotide ASO, RNAi) and translation therapy where interventions take place during translation (e.g. suppression of premature stop codon).

The first report on genetic therapy goes back to 1972, promising that genetic engineering would accomplish the reestablishment of normal gene activity. The whole idea was initially based on the introduction of genetic material to the affected cell, usually with the use of viral vectors. In vitro (ex vivo) and in vivo approaches can be used. In vitro modifications of cells extracted from the patient allows the introduction of altered cells back to the patient, while in vivo protocols include the direct delivery of genetic material to the patient. The recent and advanced understanding of the CRISPR-Cas9 (Clustered Regularly Interspersed Short Palindromic Repeats-Cas9) technology is expected to facilitate locus-specific genetic engineering modifications. The ability to create multiple breakages along different DNA loci, combined with selective repair procedures, presents an ideal tool for removing faulty parts and introducing corrective ones. For example, in trinucleotide repeat expansion disorders like Huntington Disease, the potential ability to safely remove the expansion may allow the gene to retrieve its normal size and action.

Genetically-based therapeutic approaches (drugs/formulas) have already received approval and are being used in the management of Duchenne

Muscular Dystrophy (DMD), Cystic Fibrosis (CF) and Spinal Muscular Atrophy (SMA). All approaches are genotype-driven and target either a certain type of variants (e.g. nonsense mutations) or specific pathogenic variants like CFTR: F508del in Cystic Fibrosis patients or an RNA molecule (use of ASO). Nonsense mutations are nucleotide changes introducing a premature stop codon leading to the production of non-functional truncated proteins. The existing laboratory approaches use a bioactive molecule which interacts with the ribosome to overlook the premature stop codon, thus allowing the synthesis of full-length proteins. Previous successful applications point to children, adolescents and adults with genetic rare diseases like Duchenne Muscular Dystrophy. In the case of Cystic Fibrosis, besides nonsense mutations, pathogenic variants lead to either no protein molecules and molecules with no trafficking activity (groups I&II) or protein molecules with reduced activity (groups III, IV, V and VI). CFTR modulators, potentiators (that increase gating of existing CFTR channels) and correctors (that increase the number and function of CFTR channels) are developed to repair the protein defect according to the causative variant. AntiSense Oligonucleotides are chemically modified synthetic regions (20-22 nucleotides), homologous to target RNA molecules under regulation. For example, an ASO molecule is used to induce correct splicing of SMN2 exon 7 and activate this SMN2 pseudogene in SMA patients where an exon 7 (and/or 8) deletion or SMN1 point mutations abolish the production of SMN protein. Gene therapy is also approved for SMA and includes the viral delivery of intact SMN1 to the patient.

To this end, a new term, “theragnostics/theratyping”, may be used to describe this unique field of medicine where a variety of approaches are used to reassure a definite diagnosis —in this case a pathogenic genetic variant—, for which a specific targeted therapeutic approach will be developed.

The impact of NGS is very important and highlights its game-changer character in the field of genetics. Unlimited canvassing options provided by testing more and more genetic components of the human genome (whole exome, whole genome, mitochondrial DNA) help resolve an increasing number of disorders and set solid foundations for the

advance of genetically-based therapies. Developing appropriate scientific networks for the dissemination of novel and interesting findings which may influence therapy selection, efficacy and safety and compiling patient registries that respect the General Data Protection Regulation (GDPR) will be the starting point for moving personalized treatment and other medicine options into clinical practice.

Patient or disease registries are organized systems where uniform data (including disease characteristics, symptoms, and dramatically increasing genetic variants) are recorded at national or international level. The establishment of registries assures uniformity of protocols and methods and helps facilitate collaborations between registry coordinators (scientists, patient associations, institutions) and pharmaceutical companies or state mechanisms. In the same context, biobanks (biorepositories where mainly human biological samples are stored for research) are also important for precision medicine. Samples in biobanks may become available to multiple researchers around the world and allow access to information arising from studying patients individually and as a whole. Although at the beginning biobanks were small university-based structures developed for specific projects, in the last few years they have evolved to population-based, institutional and even virtual ones to facilitate processing, storage and distribution of biospecimens and associated data. Biobanks contribute to the study of life-threatening diseases and are expected to improve prevention, diagnosis and treatment of serious diseases.

3. Genomic Newborn Screening

The incorporation of genome-wide (whole exome or genome) sequencing into a public health program or population-based newborn screening, soon after birth aims to identify infants with treatable conditions before they present clinically, prevent many more serious early-onset diseases or suffer irreversible damage, than conventional clinical and laboratory methods today.

The current public health programs for newborn screening is performed for selected conditions as a metabolic or biochemical analysis. The recommendation guidelines from the ACMG for newborn screening differ between countries regarding to which conditions and how many diseases are tested for.

Recent research has demonstrated that genomic technology such as genome-wide (whole genome or exome) sequencing, can identify genetic causes of rare paediatric diseases much more effectively than other conventional clinical and laboratory methods. Furthermore, genome-wide sequencing could, at least in theory, be used in newborn screening to identify many more serious health conditions.

There are some recommendations regarding the use of whole exome or genome, for population-based newborn screening which are:

1. Newborn screening by any method, including genomic testing, if adopted as a public health program, should be equally available and accessible to every infant born.

2. Interpretation of genomic newborn screening results requires very good knowledge of the normal (benign) variants, as well as of the pathogenic variants of every gene tested. Genomic newborn screening programs should, therefore, make population-specific allele frequencies of every gene included in the program, publicly available in a freely-accessible database. The functional consequences (benign, pathogenic, or VUS) of each allele should also be made available.

3. Publicly-funded universal newborn screening by genomic methods should be limited to diseases that can be diagnosed in the newborn period and effectively treated or prevented in childhood.

4. If population-based genomic newborn screening is introduced, it should only be offered when it includes appropriate therapeutic

interventions, clinical follow-up, genetic counselling, quality assurance, public and professional education.

5. Newborn screening by next-generation sequencing or other genomic methods should only be considered as an add-on to current first-line screening programs.

6. Current newborn screening should not be replaced by next generation sequencing or other genomic methods for any disease unless the genomic technology has equal or better sensitivity and specificity for the disease.

7. Our understanding and ability to interpret genomic variants does not justify at present the use of whole genome or exome sequencing in population-based newborn screening. Research is needed to demonstrate the clinical utility, the cost-effectiveness of sequencing, the existing health policy and ethical issues, before the use of sequencing panels is implemented for newborn screening.

Therefore, when we have to treat newborns with serious and complex symptomatology in ICU and the differential diagnosis is very important, we should consider the NGS analysis.

It is known that 3-5% of newborns will be born with a major birth defect and more than 20% of infant deaths are caused by genetic conditions. An NGS panel for the earliest and fastest diagnosis of critically ill newborns, particularly in ICU, is very important for the optimal therapeutic interventions as one third of all babies and children admitted to the ICU have a genetic disorder.

A newborn or child admitted to ICU and presenting hypotonia, hepatosplenomegaly, failure to thrive, heart abnormality, and respiratory insufficiency should have an NGS panel. The main diseases which can be covered by NGS are: cardiac, endocrinology,

gastroenterology, genitourinary, hematology, metabolic, neurology, skeletal and skin abnormalities.

4. Family and society

Genetic syndromes cause mild to severe intellectual disability, various mobility impairments (hypotonia or spasticity), many mental problems, as well as behavioural problems with autism tendencies. Moreover, certain genetic abnormalities include predisposition to malignancies. State support for the adult life of children with genetic syndromes is null. It is worth mentioning that several children could be more functional if they were provided with parallel educational/vocational support and assisted living.

The fact that we are ignorant of genetic syndromes until we cope with them personally –even though it is a matter that undeniably concerns us all– is a paradox. A high percentage of them occur randomly (de novo). Prenatal screening tests for the detection of genetic abnormalities cannot cover the entire spectrum of genetic abnormalities, at least not up till now.

International bibliography and everyday experience at a global level assure us that many people with special needs accomplish tasks that "healthy" people consider difficult or impossible. The positive attitude of the environment (family, school, society) significantly contributes to the psycho-emotional evolution, autonomy and social adaptation of those children.

Family plays a leading role in the smooth development and social integration of children with or without special needs. However, the families of children with special needs confront many-sided and complex problems that become even more complicated due to the lack of social welfare.

It is evident that the negative attitude of a society (in whole or in part) towards people with special needs makes it significantly more difficult for them to adapt to this society. In many cases, this results in isolation,

aggression and even complete rejection or non-acceptance of the problem by the patient.

All families having children with genetic diseases get in touch with doctors from many disciplines every day. Children with genetic diseases undergo various operations, receive medication and follow countless educational therapies that usually are partly covered by social security funds.

Comprehensive information and awareness-raising at public and state level, combined with radical changes in education, will contribute to the rehabilitation and autonomy of people with special needs.

Social awareness and awakening embraces associations and societies that stand by people with genetic diseases.

Patient associations and societies aim at providing scientific, counselling and psychological support to parents and children. Some important and necessary actions are summarised as follows:

- Free clinical examination of patients for prompt diagnosis, monitoring, prognosis, prevention and prenatal screening.
- Advocating people with rare genetic diseases.
- Providing financial support to families as regards expensive laboratory examinations (DNA molecular analyses).
- Organising symposia in Clinical Genetics with international participation, as well as annual information days about current scientific/medical developments and achievements.
- Organising events about special education in cooperation with municipalities and schools.
- Free systematic assessments for children by respected therapists (speech therapists, occupational therapists, psychologists).
- Organising parent support groups in the presence of an experienced psychologist-coordinator.
- Participation of children and people with genetic diseases in cultural, artistic or athletic events (Special Olympics).
- Publishing information brochures (also via e-mail).
- Establishing a communication network with other associations.

- Physical participation in annual pan-European conferences in Genetics, as well as in Eurordis.
- Broadening relations with ministries, the Media, agencies etc.

It is crucial to establish model daycare and educational centres to promote assisted living (for children above 3 years of age).

These model centres help people with rare genetic diseases as well as their families, providing further opportunities for creative activities, speech therapy, physiotherapy, occupational therapy and psychological support. Thus, they represent a milestone not only for the rehabilitation of people with rare genetic diseases, but also for their social and vocational integration in general.

Supporting children and people with rare genetic diseases is a matter that concerns society as a whole, and not only the affected families. Examples of such genetic diseases are the following: Angelman, Prader-Willi, Noonan, Williams, Rett, Di George, Soto's, Silver-Russell, Apert, Crouzon, Kabuki, Stickler, CHARGE, Lowe, various skeletal dysplasias, chromosomal abnormalities such as Down syndrome, Turner syndrome, Klinefelter syndrome and more. Information, social and financial support and scientific guidance regarding the peculiarities of these diseases are starting principles indicating the auspicious evolution of patient associations in these demanding times.

4.1. Emotional and behavioural reactions of children having siblings with genetic diseases and developmental disorders

The relationship of two people is characterised based on the type of interaction they have. More specifically, hierarchical-complementary relationships are the ones with uneven distribution of knowledge and power (e.g. parent-child and teacher-student relationships), whereas mutual-equal relationships are characterised by an uneven distribution of knowledge and power, such as friendship.

Sibling relationships are special, since they are a combination of complementary and mutual types of relationships (Hinde, 1979). These last longer than any other relationship (inside or outside family) and are characterised by intense positive, negative or even ambivalent feelings. The sibling bond is governed by two main characteristics: proximity and confrontation (Smith & Hart 2011; McGuire, 1996).

It is very important that a sibling relationship has positive foundations that positively affect its evolution. In early and middle childhood, siblings spend a large part of the day together and create a very strong bond. Though this relationship, siblings learn and develop cognitive, emotional and social skills; at the same time, self-regulation and the sense of belonging are promoted (Milevsky, 2016; Brewton, 2012; Smith & Hart, 2011; Orsmond & Seltzer, 2007).

The quality of the relationship among siblings is affected by their differences and family dynamics. In particular, gender plays a decisive role in proximity, while birth order is also important. Older siblings assume the role of the “teacher” and usually have the upper hand, whereas the younger ones benefit more when they develop positive relationships. Lastly, a bigger age difference strengthens proximity and reduces confrontation.

In terms of interaction, siblings spend much of their time together. The ability to play depends on the selection of the right game. It may be difficult to find a common activity due to possible social deficits.

Most sibling relationships are characterised as positive and fulfilling. According to surveys based on statements made by children, there is less confrontation between siblings when one of them has disabilities compared to cases where the development of both children is normal. On the contrary, observation-based research indicates no difference between the two scenarios.

Early studies used to show exclusively negative effects when one child had disabilities. These studies were focusing on possible financial burdens, limited opportunities for recreational activities, shame and peer ostracism, as well as on the strong sense of responsibility towards a child with deficits.

Recent studies question the above position, as they acknowledge both negative and positive aspects of such a relationship. Positive aspects include increased tolerance towards diversity, empathy, higher maturity compared to peers, and meaningful appreciation of the child's health and abilities.

In several cases, the above can lead to increased stress levels for a child with normal development. This is expressed through withdrawal from family, friends and usual activities, aggressive and judgemental behaviour, persistent effort to be the "good guy", signs of perfectionism and psychosomatic symptoms.

To prevent and manage negative reactions from children, it is essential that the children communicate and express their feelings, as well as that they are provided with honest and age-appropriate information regarding this matter.

4.2. Autism through the eyes of a parent

People with Autism Spectrum Disorder (ASD) have trouble with social communication, stereotyped and repetitive behaviours, activities and interests. Autistic people are characterised by their peers as eccentric, difficult and unsociable.

Autism Spectrum Disorder (ASD) is caused by genetic and metabolic syndromes, as well as perinatal, external, neuroanatomical, neurochemical, neurohormonal and immunological factors.

The investigation of each case of autism is important since autism spectrum virtually represents a group of symptoms and is not considered a cause itself. Therefore, details about the case form a picture of the child's abilities, which contributes to a safer prognosis. This helps parents in terms of stress relief and making proper decisions for the future.

Autistic people themselves define autism as a different way of brain functioning, thinking, expressing feelings and perceiving the world; they don't consider it a mental disorder.

Ideally, an early prognosis is performed by a paediatrician, teacher or parent in children of 3 to 4 years old or in the early school years. The paediatrician knows the child and its environment very well. Using a holistic approach, they can observe the overall behaviour of the child compared to normal development and detect hearing or eye problems or other autism-related problems via m-chat. After the first stages of diagnosis, the paediatrician can refer the child to a multidisciplinary team for a more comprehensive assessment and, when necessary, early intervention.

Early intervention is crucial since it capitalises on brain plasticity and achieves early preparation for school integration. This procedure prevents the establishment of pathological behaviours.

There are various treatment approaches for autism in children. These include occupational therapy, speech therapy, pharmacotherapy, Applied Behaviour Analysis (ABA), social stories, TEACCH, PECS, sensory integration, music therapy, Cognitive Behavioural Drama (CBD), the developmental movement method (Sherborne) and therapeutic horse riding.

Parents consider autism a torture or even a challenge to adopt a different perspective of life itself. In practice, following autism diagnosis in a child, parents have to deal with a child that is now stigmatised and excluded from the social fabric.

At first, parents are completely ignorant of the nature of the disorder due to the lack of information from the State and even experts. Therefore, they are required to cope with various challenges, such as how to shape their relationship with the child without undermining it, how to handle the relationship with the spouse, siblings and social circle, as well as how to deal with social exclusion. Combined with the lack of information, these concerns lead parents to look for answers in questionable web pages, without being able to handle this information.

Diagnosing and managing autism translates into a loss that represents the end of a known situation and the transition to something unknown and frightening. Inevitably, loss leads to grief, which goes through 5 stages (Elisabeth Kübler-Ross): denial, anger, bargaining,

depression, and acceptance. Throughout these stages, it is crucial to support and encourage parents, showing respect to their feelings.

Protecting the parent's mental health is the cornerstone of successfully managing the situation and ensuring long-term results. This is achieved at first though an individual psychotherapy programme, then through expert guidance and counselling. Relevant parenting education programmes (Early Bird, Cygnet), as well as parent groups and associations, also provide help.

A good therapist is a trained professional eager to help. The therapist acknowledges and respects the limits of an autistic child, preventing tantrums during the session, and personalises their treatment approaches according to each child's needs. They can objectively assess the appropriateness of an activity and redefine short-term and long-term goals. Lastly, they do not reciprocate anger; instead, they adopt a soothing and calming approach.

Autistic children monopolise parents (especially during the first years after diagnosis) and often become manipulative and very demanding. On their side, parents have a hard time setting boundaries and, in many cases, unconsciously put pressure on their environment.

The education of children is a source of anxiety for parents. A scientific methodology is not always required, and it is often enough to take actions completely tailored to the child, e.g. whispering in the ear, pointing the index finger or visualising information. Parents have to respect stereotypity and different way of thinking. Meanwhile, the child's creative thinking and flexibility are of paramount meaning. In terms of education, the boundaries are set by the parent together with the child. Parents should not think that this way they will spoil the child; they are supposed to aim at discovering, boosting, nurturing and encouraging the child's strengths. This will satisfy the child and give great pleasure to the parent. Lastly, the child's school environment varies depending on severity. Integration classes, parallel support and special schools are all possible options.

Autism means that people coming into contact with the child have to alter their natural way of communicating.

Meanwhile, the couple is faced with daily tension, and the intractable problems arising from Autism Spectrum Disorder gradually shake the couple's relationship, leading to separation or divorce. Under such conditions, it is usually the man that gives up first, while the mother is left alone with an unbearable burden.

It is observed that siblings assume the role of the caregiver, taking on disproportionate responsibilities. This deprives them of their childhood and spontaneity. In an attempt to draw the parent's attention, they often imitate pathological/autistic behaviours. Growing up in such a family, they cannot spend quality time with their parents due to the long-term treatment of the autistic child, and sometimes they are clearly sidelined. As they mature, the siblings of an autistic child tend to opt for professions related to taking care of people with special needs (psychologists, special education teachers, speech therapists etc.).

4.3. After-school prospects for people with disabilities

Disability is defined as a complex and varying phenomenon resulting from the interplay of personal characteristics and the characteristics of the environment in which one lives. According to this definition, people with physical defects may or may not experience disability in an environment, depending on the barriers and aids this environment incorporates (*National Confederation of Disabled People of Greece*, *2005*).

The medical model defines disability as a physical, mental, sensory or psychological "deviation" from what is considered "normal"; it is a "dysfunction" caused by a disease, accident or other medical factors. The medical model identifies philanthropy and provision of benefits as the solution (*National Confederation of Disabled People of Greece*, *2005*). At the same time, it promotes "separation" practices such as institutionalisation and exclusion of people with special needs from any domain of social life, including employment (*Gottlieb, Myhill, Blanck, 2010*).

The social model acknowledges disability as a problem that is caused by society itself and is not treated in medical terms. To solve this problem, people with disabilities need to assert human rights and apply for jobs. Meanwhile, efforts to improve the qualifications and skills of people with disabilities and to change the employers' negative attitude towards the abilities and skills of these people are of great importance (Gottlieb et al., 2010).

People with disabilities can enter the workforce through protected, assisted, or adapted employment. Protected employment includes specially designed workshops for people with disabilities. These conditions are characterised as closed market conditions (Lenhart, 2000). Assisted employment is based on the official paid employment contract and social security, under the method of "systematic guidance" (Armpounioti, 2003). Adapted employment focuses on the individual choice, needs and qualifications of the disabled person, with reasonable adjustments (Gottlieb et al., 2010).

The transition from protected employment to assisted employment varied substantially depending on country, and the implementation required generous funding to support structures, expansion of employment agencies, employment subsidies, and retraining of the employees in new techniques and skills (Cummins and Lau, 2003). Experience showed that when the transition programme begins at an early age, this better ensures that the person will stay in the workforce and feel satisfied with the support they receive (Cimera, Burgess, Bedessem, 2010).

Another factor dynamically affecting the decisions and life of employees with disabilities is their family. In particular, successful transition requires the inclusion of family in planning, since their expectations and the support they offer affect their children's behaviour and performance (Neece, Kraemer, Blacher, 2009; Boehm, Carter, Taylor, 2015).

The employers' role in the effort concerning the inclusion of people with disabilities in the labour market is equally important. Factors such as the characteristics of the employer's and candidate's personalities, the employer's beliefs and attitude towards disability, and the

recruitment process itself affect the final decision making (Zappella, 2015). A survey conducted by the Greek Manpower Employment Organization shows that prejudices are reduced when employers hire people with disabilities in their business (Logaras, 2013).

Reforming the education policy can significantly boost smoother and more efficient inclusion of people with disabilities in the workforce. In particular, customised education programmes, training, provision for a transitional body, and occupational integration planning for people with disabilities can help and offer a holistic approach to disability.

The implementation of the above requires internships in work environments outside school, support services for all parties (follow-up), continuous assessment at each stage, as well as linking education to the labour market.

4.4. Sexuality and sex education of people with special needs

The World Health Organization defines sexuality as an integral part of personality; it is a fundamental need and aspect of a person and cannot be separated from the other aspects of life.

Sexuality is associated with the deep human need for being likeable and accepted, giving and receiving affection, feeling worthy, being appealing and able to share thoughts and feelings, and includes sex, gender identities and roles, sexual orientation, eroticism, pleasure, intimacy and reproduction. Meanwhile, it is affected by the interplay of biological, psychological, social, financial, political and cultural factors.

The sexual behaviour of people with disabilities is shaped by the attitude, values and norms of family, society and peers, the child's relationship with parents, parental control, as well as the quality of the communication with parents. The best parent-child relationships are associated with non-early sexual intercourse, less frequent intercourse and fewer sexual partners.

The psychosexual development is the same for everyone, including people with special needs. However, social perception did not always allow the normal expression of sexuality by people with special needs. Initially, these people were institutionalised in order to be “taken care” of. On this basis, any attempt to approach and communicate with the opposite sex was severely punished. This imposed restrictions in the expression of sexuality. Afterwards, deinstitutionalisation of people with disabilities was implemented within the framework of human rights protection. Meanwhile, the legislative framework was amended in order to facilitate non-voluntary sterilisation for the mentally disabled. In the last few years, there has been an increased research interest, and sex education for people with disabilities is a reality.

Even nowadays, there are still obstacles. These obstacles are mainly imposed by wrong family and society perceptions, for example the idea that people with special needs are asexual, sexual development is a risk source, sex education will increase sexual behaviour and people with disabilities are incapable of making decisions. The lack of education programmes for parents/caregivers and disabled people themselves presents a still existing important barrier.

People with disabilities have needs and concerns related to sexuality. More specifically, they are often unable to determine what is considered acceptable behaviour in a relationship. Moreover, there are concerns about finding the balance between protecting someone against getting hurt by a relationship and offering them the autonomy to make decisions. Managing the feelings of people with disabilities in also an important factor.

These people are in need of sexual relationships, intimacy and friendship. Meanwhile, it is necessary to educate people regarding sexuality and to support them in order to start and maintain a relationship, without violating their privacy. People with disabilities want to feel that their wishes and needs are heard, taken into consideration, and respected (Brown & McCann, 2018).

Parents and caregivers of people with special needs seem to lack confidence and ability to discuss matters pertaining to sexuality. They also seem unable to manage the distance between sexual expression of

children and other, more childlike traits in their behaviour. Parents/caregivers have a strongly ambivalent attitude towards sexuality; on one hand, they want to let children fulfil their desires, but they are protective of them at the same time. The proposed solution is education through separate programmes for parents/caregivers and children, as well as through group programmes (Brown & McCann, 2019).

Sex education is a life-long learning process that relates to the knowledge one has about themselves and their development as a social and sexual being. Objectives include providing information, developing values and interpersonal skills, strengthening responsibility and reducing risks. Main education topics concern information pertaining to the physiology of organisms, sexual intercourse, relationships, self-consciousness and safety.