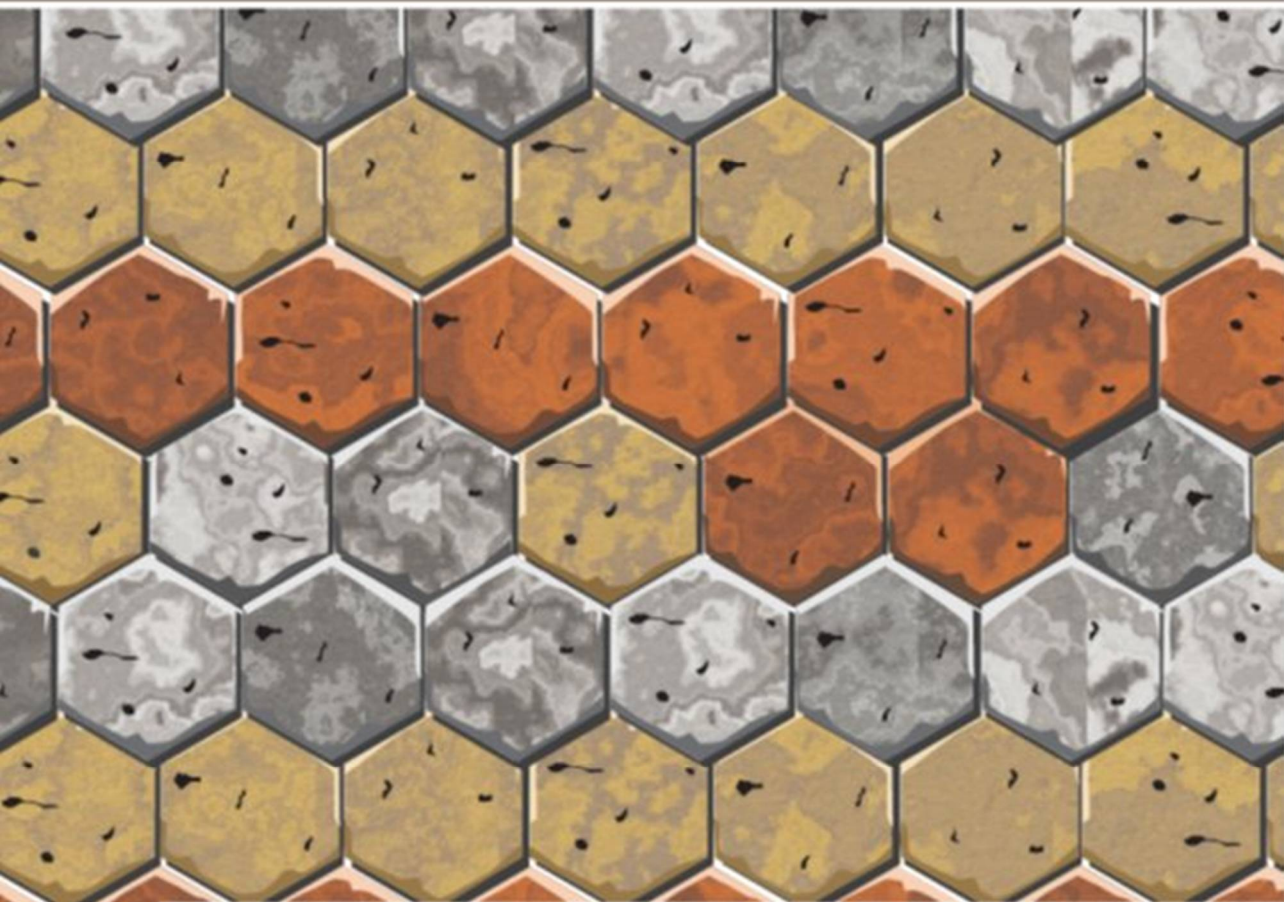


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A MOTHER'S EXPERIENCE OF FACING THE DIAGNOSIS OF PALLISTER KILLIAN SYNDROME

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Abstract

Pallister Killian syndrome is a rare genetic disease, the 12p chromosome is defective. People with Pallister Killian syndrome have problems in all areas of functioning. Intellectual deficits and associated diseases are common. Just as the deficits are permanent, so is the help that a person with Pallister Killian syndrome will need throughout life. Informing the parents about the diagnosis often means giving them less favourable news. Parents receive information about the diagnosis from medical professionals, and then they have to familiarise themselves with the disease, confront it and live with it. The article presents how a girl's mother coped with the diagnosis of Pallister Killian syndrome, who offered her the most support and what she missed the most in the process of coping with the diagnosis. The research found that in this early period, systemic support from various health service and treatment providers, as well as help in asserting the rights she was entitled to due to her child's diagnosis, were most missed by the mother.

Key words: Pallister Killian syndrome, learning about the diagnosis

Introduction

Pallister Killian syndrome (PKS) is a rare genetic disease characterised by individuals having a portion of completely normal twelfth chromosomes, while other chromosomes have tetrasomy on the p arm (Al Shehhi et al., 2019). The prevalence of persons with PKS is 5.1 per 1,000,000 live births (Wu et al., 2021), there are 300 to 500 persons with this diagnosis in the world. Despite the

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awareness of PKS, the diagnosis is still often made late (Internet 2). A person's age is a key factor when diagnosing PKS. More mosaicism is present in younger people, which is why it is easier to make a diagnosis – the percentage of mosaicism decreases with age (Toydemir et al., 2020).

Numerous clinical signs can be observed in people with PKS, which manifest themselves in a very different spectrum (Toydemir et al., 2020). Newborns may have a markedly large head (Jamuar et al., 2012) and a low Apgar score (Internet 2). Common congenital abnormalities are characteristic. Various malformations are often present, e.g. kidney dysplasticity and congenital heart defects. The mentioned problems can be so pronounced that they overshadow the remaining features of PKS. This results in a later diagnosis, as it is inadvertently overlooked due to all the other problems (Jamuar et al., 2012). It is characterised by low muscle tone (hypotonia), which can affect the appearance of more notable motor deficits. Children reach developmental milestones later, and infants may be developmentally delayed (Internet 2). Developmental delay is visible very early after birth (Fetta et al., 2022). People with PKS may be mobility impaired and have congenital or acquired defects of the locomotor system (Internet 2). In the first months of life, there is a pronounced hairiness of the forehead and ears (Arghir et al., 2021), but later the hairiness of the scalp is worse, especially in the frontal-temporal area. People with PKS have sparse eyebrows and eyelashes (Jamuar et al., 2012). Skin changes (hyperpigmentation) and joint contractures are also common (Al Shehhi et al., 2019). Joint contractures occur mainly in adults with PKS, and they affect the mobility of the joints, principally the elbows (Internet 2). It is the pigmentary changes on the skin and the unusual hairiness of the scalp that should lead diagnosticians to consider the diagnosis of PKS (Jamuar et al., 2012).

Speech is often less developed or even completely absent in people with PKS. Vision and hearing are also impaired (Internet 1). Research shows that as many as three quarters of children with PKS who are older than 18 months do not speak (Internet 2). An autistic spectrum disorder often accompanies PKS. This further hinders the development of speech and language as well as a wider communication range (Kostanecka et al., 2012). Only a few children are strong in the social field. Most do not spontaneously establish contact with the environment. Establishing eye contact is also absent (Internet 2).

Epilepsy is present in 39-59% of persons with a confirmed diagnosis of PKS (Giordano et al., 2012). Epilepsy usually occurs during the first four years of life (Alqhatani et al., 2019). The most common forms of epileptic seizures in people with PKS are focal seizures, epileptic spasms, and myoclonic seizures (Ricci et al., 2019). People with PKS have problems processing sensory stimuli, they can overreact to them or not react strongly enough.

The association of deficits with the primary PKS causes additional complications and greatly hinders learning processes and the acquisition of basic academic and practical knowledge. As people age, behavioural problems increase. These problems are greater in people with communication and intellectual deficits (Internet 2).

An accompanying sign of PKS is the presence of intellectual deficits, which are manifested in varying degrees and intensities.

Just as the deficits are permanent, so is the help that a person with PKS will need in life.

Getting to know and confronting parents with the diagnosis

When parents find out that their child is "different", their world changes dramatically, and the foundation of the family bond can be shaken. Their everyday life begins to change. Making a diagnosis often means that several visits to different specialists are necessary. These are followed by taking the child to various medical and therapeutic treatments. Caring for a child with the rare diagnosis requires more time for basic activities than caring for a healthy child (Kolemen et al., 2021). Because of everything that changes in their lives, parents are often the first to need help (Restoux, 2010). The diagnosis most often answers questions about the health conditions (David, 2015). Making the diagnosis significantly changes the life of the patient and his/her loved ones (Prevc and Zrnec, 2016). The role of the environment in this is to understand them and provide them with support. Parents get support from relatives, family and friends (Restoux, 2010).

Usually, the diagnosis of a disease or condition is unwanted information. No parent wants to hear that their child has a condition that will affect their quality of life. The diagnosis also affects the quality of life of the parent and the entire family. On the other hand, it is the diagnosis that enables, for example, the beginning of treatment, it is easier to regularly access educational, therapeutic, social and other services in the field of rehabilitation (David, 2015). Early diagnosis can lead to better outcomes later in life. Children who are diagnosed before the age of four have better opportunities for

various measures, and by the time they enter school, they are more successful in the verbal and intellectual fields than children who are diagnosed later. Children who are diagnosed early are supposed to need less support and help later. Along with the desire to diagnose as early as possible, it is also necessary to provide adequate help and support to parents (Rabba et al., 2019).

Some parents do not wish to find out the child's diagnosis immediately after birth, especially in cases when it is permanent, as this allows them to have at least some personal time with the newborn without the diagnosis (David, 2015). There is probably no right time or a right way to deliver bad news (Dodič, 2016), and delivering the diagnosis is often just that - bad news.

Parents have numerous questions during this period. They often do not get the right and/or timely answers. They need support in finding information in order to provide the child with appropriate and high-quality treatment (Restoux, 2010). It is the duty of healthcare professionals to present all the details about the disease to parents (Rancigaj Gajšek, 2018). Parents have the right to an accurate answer about the child's condition. Therefore, it is important that doctors are honest when answering questions (Dodič, 2016).

When communicating a diagnosis, the most important way of communication is at the beginning - this forms the relationship between the doctor and the patient, or his/her relatives. You only learn bad news once, and the entire coping process depends on it (Ranchigaj Gajšek, 2018). The diagnosis causes parents pain, which they cannot be spared. It is important to be empathetic when communicating, we must pay attention to the choice of words with which we tell the parents the diagnosis (Dodič, 2016). The way the diagnosis is communicated and the time it takes to support the parents who are faced with their child's diagnosis are often referred to as a "black hole". During this time, parents cope with the diagnosis and sometimes deny what they see (Duma et al., 2021).

Insufficient knowledge of rare diseases can also be a challenge for health professionals involved in diagnosing people. Despite modern and advanced molecular genetic diagnostic systems, it is not possible to diagnose all rare genetic diseases. Waiting for a diagnosis can be a very challenging time for children and their parents. Such children often visit several specialist doctors and it takes several years to get a proper diagnosis (Kolemen et al., 2021).

When searching for the answers to their questions, parents can be helped by health professionals of various profiles and specialties, pedagogues, families who have children with the same or similar

diagnosis, (self-)help associations and others. Parents can also find a lot of information online. When searching for information online, they must be critical of the offered content. The parents who are not proficient in a foreign language, face additional problems when searching for answers, as it is more difficult for them to obtain information about their child's diagnosis.

The time when parents find out about their child's diagnosis determines the kind of support they need to cope with the situation. Regardless of the time of making the diagnosis, the family plays a big role in coping with the diagnosis. Immediately after receiving the diagnosis, the support of all family members is important. Mothers rely primarily on fathers for support. Grandmothers often provide great support, helping parents take care of the child and also providing them with emotional assistance. Some parents mention that the support of their brothers and sisters, who are like a third parent of their child, also means a lot to them (Bruns and Foerster, 2011). After receiving the diagnosis, the parents are repeatedly subjected to a rollercoaster of different emotions. Among these there are frequent fluctuations between relief and shock (Downes et al., 2021). Parents often experience stress during this period. In addition to stress, some also experience despair and self-blame, which affect their mental health, the quality of their lives and the life of the entire family (Rabba et al., 2019). All the emotions experienced by parents upon receiving the diagnosis also have an impact on the partner relationship - both positive and negative (Downes et al., 2021).

Aim and goal

The aim of the research is to establish which key factors influenced the mother of the girl with the diagnosis of PKS. We want to determine how the mother coped with the diagnosis, what helped her, what her partner's role in coping with the diagnosis was, and what the wider environment was like.

Research question

1. When did the mother find out about the child's diagnosis?
2. Who was the first to inform the mother of the diagnosis? In what way was the diagnose communicated to her?
3. What questions did the mother have when first meeting with the diagnosis?
4. Where did the mother look for answers to the questions she had?
5. Where did the mother look for help in learning about the diagnosis?
6. Where did she seek/get support when learning about the diagnosis?
7. What forms of assistance helped the mother in learning about the diagnosis of the rare disease?

Methods

Sample

The sample included the mother of the girl, who at the time of the research was the only one in Slovenia diagnosed with PKS. The mother of the girl with PKS is 43 years old. She is a preschool teacher by education and works as an assistant teacher in a kindergarten and has 15 years of work experience. She was 26 years old when the girl was born. There are two children in the family, the first-born is a girl with PKS, and the second-born is a healthy boy. Due to her daughter's diagnosis and because she takes care of her, she works part-time. For the other four hours, he receives partial compensation for lost income.

Data and analysing methods

For the purpose of the research, a questionnaire that contains 20 sets of open-ended questions was created. The questions relate to learning and dealing with the diagnosis of PKS, knowledge about PKS and observing the signs that are characteristic of this diagnosis and are also present in her child. Based on the answers, a semi-structured interview was prepared. It was used to help obtain in-depth answers to the questions. A descriptive method and the causal-non-experimental method of pedagogical research was implemented. A non-random opportunity sample that included the mother of the child with Pallister Killian syndrome was used in the research. A qualitative singular case study was conducted. It determined how the mother of the child came to learn about the diagnosis of PKS.

Results

The following is an interpretation of the answers to the questions asked. Firstly, the mother went on to describe the pregnancy, which was uneventful. The only problem she had was high blood pressure. No special features were detected in the girl at birth. The parents noticed the first signs of developmental delay when the girl was about six months old, as she showed no interest in her surroundings. At the age of nine months, the girl was referred to neuro-physiotherapy and to a developmental clinic due to developmental deficits, and later to a neurologist, and from there to a geneticist, where genetic blood tests were performed. The results were normal. At about a year and a half, the parents began to notice unusual twitches every time the girl woke up. Further tests revealed the presence of epilepsy. At the age of three, the girl underwent genetic tests again, this time from a saliva sample.

The mother learned about the diagnosis through a phone call she received from a clinical geneticist while at work. At first, she only told her that the results of the analysis revealed something, and asked her to bring the girl in for another swab sample. The mother was eager to learn what the tests showed and the clinical geneticist told her it was the Pallister Killian syndrome. Upon arriving home, the mother started researching the diagnosis online. The girl's resemblance to the photos of the children she found was considerable, so she knew the diagnosis was correct. When they came to see the clinical geneticist, she had prepared questions that she wanted to ask her.

When asked about what questions arose after learning about the girl's diagnosis, the mother said that she and her partner were mainly interested in how the girl's development would proceed, whether the girl would ever be independent, would she ever speak, what kind of medical conditions she would have problems with and what her life expectancy was. Among the features of Pallister Killian syndrome in the girl, they noticed mainly developmental delay, hypotonia, poor speech development and some dysplastic signs.

When the mother was asked where she looked for answers to the questions in order to better understand the girl's diagnosis, she answered that on the Internet. At that time she did not find any answers in her native language. The literature was only available in English. She found an American website established by the parents of children with PKS, she got some answers to her questions on a forum. Today she receives the most answers to her questions on the

social network Facebook, in a group for the parents of children with Pallister Killian syndrome, which has about 500 members.

The mother says that she and her partner did not receive outside support. They sought it mainly from each other, but they also got some from the parents of children with Pallister Killian syndrome abroad. They missed a holistic approach, someone who would keep an eye on the entire situation. To some extent, the procedure was run by a developmental clinic, but at some point, the girl was too old to continue being treated there. Now, to a greater extent, especially in the behavioural field, a child psychiatrist has taken over.

According to the mother, a clinical geneticist would be the most appropriate person to help her and her partner learn about the diagnosis of Pallister Killian syndrome. They were somehow left alone in the situation. What the mother missed the most was an institution that would take over a part of the burden of the parents of children with rare diseases. Above all, she missed help in asserting the rights that belong to them. She and her partner learned about most of these from other parents.

The mother says that she does not remember exactly how she and her partner announced the diagnosis, but they did not experience any major fears. It took some time for her to come to terms with the diagnosis, but the girl was very loved despite her developmental delays. The diagnosis did not change anything. The diagnosis gave them answers to certain questions, in a way they learned how to move forward more easily.

Discussion

With the help of research, we answered the research questions. The answer to the first research question, "When did the mother find out about the child's diagnosis?", is that the mother found out about the girl's diagnosis when the girl was little over three years old.

The answer to the second research question "Who informed the mother of the diagnosis?" In what way was she informed?" is that the diagnosis was communicated to the mother over the telephone by a clinical geneticist. A few days later, the mother and the father were informed of the diagnosis in person, at the clinical geneticist's clinic.

The answer to the third research question, "What questions did the mother have when she first met with the diagnosis?", is that the questions were mainly related to the girl's future, to the course of diseases that may be associated with Pallister Killian syndrome.

The answer to the fourth research question "Where did the mother look for answers to the questions that arose?" is that the mother

looked for answers to the questions that she had online, in articles on various foreign websites and in a group of the parents of children with Pallister Killian syndrome, which was created on the social network Facebook.

The answer to the question "Where did the mother look for help in learning about the diagnosis?", is that the mother looked for help and support mainly from her partner, and he looked for them from her. She also sought help in learning about the diagnosis online, from parents who had similar experiences. The same answer can be given to the question "Where did she look for/get support when learning about the diagnosis?", as she looked for and got it from her partner and the parents with similar experiences.

The answer to the last research question, "What forms of assistance helped the mother when she was diagnosed with the rare disease?", is that no form of systemic assistance was offered to the mother and father at the time of the diagnosis of the rare disease, e.g. the possibility of visiting a psychologist. From September 1, 2019, this area is covered by comprehensive early treatment in Slovenia.

From the interpretation of the obtained results, we note that the mother found out about the girl's diagnosis when the girl was a little over three years old (RQ1). The diagnosis was communicated to the mother over the phone by the clinical geneticist, and a few days later it was communicated to her and her partner in person, in the clinical geneticist's clinic (RQ2). Initially, the most important thing is the way the diagnosis is communicated, as it shapes the relationship between the parents and doctor (Rancigaj Gajšek, 2018).

The mother mainly had questions related to the girl's future and the course of the disease, which may be associated with Pallister Killian syndrome (RQ3). The diagnosis is the element that provides answers to questions about health conditions and enables the start of treatment and access to various treatments and services (David, 2015).

The mother looked for answers to her questions on the Internet, on various foreign websites and in the group of the parents of children with Pallister Killian syndrome (RQ4). (RQ5). Parents can obtain a lot of information online, but it's important to consider it with the right amount of criticism.

The mother sought and received support from her partner and the parents of children diagnosed with Pallister Killian syndrome (RQ6). The family plays a big role in the process of coping with the diagnosis, as it provides support to its members.

The mother and the father were not offered any form of systemic assistance during the process of learning about the diagnosis (RQ7).

Conclusion

The purpose of the research was to determine the key factors that influenced the mother's coping with her daughter's diagnosis - PKS. We found that the mother coped with the diagnosis more easily after learning about the characteristics of Pallister Killian syndrome. Knowing the characteristics somewhat calmed her down, because in this way she found out what kind of future she could expect. In the process of facing the diagnosis, she received the greatest support from her partner, as they both had to face the diagnosis. They reacted differently to the situations, as they each faced the stress of the rare disease in their own personal way.

Bodenmann named the way couples use their relationship to manage stress together "dyadic coping". "Dyadic coping" describes the way in which stressors indirectly or directly affect the relationship between parents. A partner can be directly affected by the stress his partner is experiencing. The ocean of stress is transferred verbally or nonverbally between the two partners, the partner interprets the received information and reacts to it with a positive or negative "dyadic" response. A positive response is supportive and helpful, while a negative response is hostile, superficial or just an ambivalent attempt of support (Downes et al., 2022).

The girl's parents successfully coped with the stress that their daughter's diagnosis of Pallister Killian syndrome brought to their relationship. This affected their positive attitude and the good functioning of the entire family. Due to the successful coping with the diagnosis and the adequate support from her partner, the mother did not feel any stress when announcing the diagnosis to her relatives and friends.

In the process, systematic support was what the girl's mother missed the most, e.g. healthcare workers support, assistance in asserting rights, etc. Later, this was regulated in Slovenia with the Act on Comprehensive Early Treatment of Preschool Children with Special Needs, which came into force in September 2019. The Act provides for the care and provision of various services for both the child and his family.

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