January 2022

Rare Disease Research Partners

Psychological Support at Diagnosis of a Rare Disease

A Review of the Literature

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Contents

INTRODUCTION

Approximately 6% of the population is affected by a rare disease (1). The diagnostic odyssey is significant, and patients and their families can often be left waiting years for a confirmed diagnosis. Delays in diagnosis can have medical consequences such as delayed treatment, unnecessary tests, and psychological stress for the family (1). Even after diagnosis there can be a lack of both information, and healthcare specialists with the knowledge to support those affected (2, 3). Caregivers are undoubtedly additional victims, often taking on this role under sudden and extreme circumstances, with minimal preparation and little guidance and support from healthcare systems (4), and the psychological impact on them, affects other members of the family (5). The danger is that if the caregiver burden becomes too great and their well-being is compromised, they will no longer be able to provide the vital care needed by the child (4).

The diagnosis of a rare disease is often difficult to understand well immediately and will generate many questions and uncertainties (6). The moment of diagnosis of a rare disease is psychologically complex. At the point of diagnosis parents are often torn between feelings of relief, and feelings of sorrow and/or guilt (7). A diagnosis could be perceived as the first step towards treatment, a way of reducing a child's suffering, or as a turning point that could enable families to start adjusting to their new normality (8). Stress at this time is often manifested by anxiety, uncertainty, worry, fear, frustration, grief, powerlessness, shock, or denial. Parents often feel overwhelmed with sadness, feelings of vulnerability, suffer from anticipatory loss, blame, confusion, disbelief, dismay, helplessness, insecurity, and perceive a lack of control. (9). Those who have endured some degree of diagnostic delay often initially experience more relief (8).

The high levels of reported stress in parents and carers of those with rare and complex diseases suggest an unmet need for more formalised psychological support from mental health professionals, counsellors, or peer support groups (1). The availability of professional counselling with minimal delay might be helpful for those affected to regain and restore confidence, and to build effective coping strategies for both the current situation and future adversities.

This report is based on evidence collected through reviewing published literature systematically. It focuses on the psychological impact actions and interventions have around the time of the diagnosis of a rare disease and seeks to elaborate on what might make a difference to those affected, and what might be done to improve outcomes in the longer-term through improved psychological support at this crucial time. The literature reviewed focuses on diagnoses of rare diseases made in infancy and early childhood, although the findings and their implications are likely to be more widely applicable. While there are significant psychological sequalae of living with a rare and complex disease, the focus is on the support that could be built around the crucial time of the diagnosis, as this appears to lay the foundations of much the family experiences, and how they perceive and react to other events in their journey.

METHODS

In order to source relevant information, the keywords “psychological support AND rare disease” were used to searched PubMed. The search strings incorporated free text key words and Medical Subject Heading (MeSH) (Table 1).

Literature Search Using PubMed

Keyword	Search strings	Number of articles
Psychological support AND rare disease	("psychologic"[All Fields] OR "psychological"[All Fields] OR "psychologically"[All Fields] OR "psychologization"[All Fields] OR "psychologized"[All Fields] OR "psychologizing"[All Fields]) AND ("support"[All Fields] OR "support s"[All Fields] OR "supported"[All Fields] OR "supporter"[All Fields] OR "supporter s"[All Fields] OR "supporters"[All Fields] OR "supporting"[All Fields] OR "supportive"[All Fields] OR "supportiveness"[All Fields] OR "supports"[All Fields]) AND ("rare diseases"[MeSH Terms] OR ("rare"[All Fields] AND "diseases"[All Fields]) OR "rare diseases"[All Fields] OR ("rare"[All Fields] AND "disease"[All Fields]) OR "rare disease"[All Fields])	724

Table 1. PubMed literature search

Screening

Types of studies

We did not exclude any study design, nor were there exclusions based on sample size, length of follow-up, or country.

Selection of studies

All references identified through the search strategy were downloaded into Endnote Version X9. Two reviewers independently screened the references for potential inclusion based on title and abstract. Reviewers were not blinded to study author or journal. A third reviewer was planned to be used to mediate any disagreements until consensus was reached, however, this was not required. We contacted the study authors directly for any papers with insufficient detail to determine eligibility. We obtained the full texts of studies deemed potentially eligible for inclusion, and two reviewers independently assessed them for eligibility.

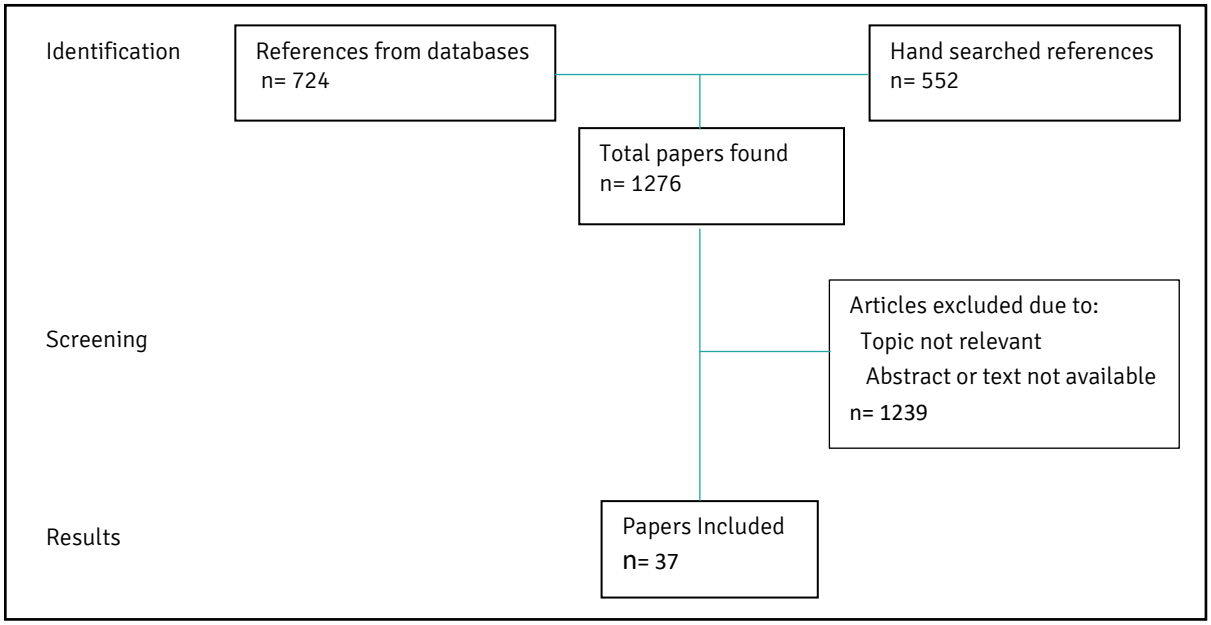
Those studies deemed as ineligible were recorded with reasons for ineligibility. We created a flow chart following the PRISMA protocol to illustrate this process (10).

Data Extraction

Two reviewers independently extracted data onto a piloted data extraction form developed for this review, with discrepancies resolved via consensus discussion.

Flow Diagram

A total of 1276 papers were identified and screened. Of these papers, 1239 were excluded due to irrelevance or unavailability of the text, and the remaining 37 papers were taken forward for inclusion in this review (1, 3, 7-9, 11-42).



WHAT HAS A NEGATIVE IMPACT AROUND THE TIME OF DIAGNOSIS

At diagnosis, families affected by a rare and complex disease are best characterised as emotionally and financially stressed, with the costs of caring for a child with a rare disease being significant. Families may need to pay for travel costs, day-to-day care and for adaptations to the family home. They desire better communication and coordination of care, feel frustrated with the diagnostic process, they value improved access to information, and require greater social and psychological support (25). Many families feel that their experience is characterised by delayed diagnosis, inadequate information, and lack of psychological support (1).

There are a wide range of domains of supportive needs for parents caring for a child with a rare disease, they include practical, financial, spiritual, social, psychological, informational, emotional, and physical needs (9). Their psychological needs are often described by parents as manifesting as feelings of uselessness, powerlessness, and helplessness in their care for their child with a rare disease (9). These feelings are compounded by endless worry about their child's prognosis and the progressive nature of the condition (33).

Prolonged Diagnostic Odyssey

There is a large and ongoing impact of living with symptoms and needs, without a diagnosis. The vast majority of parents describe the period from the appearance of first symptoms to diagnosis as the most difficult and psychologically painful (36). This delay in diagnosis causes strong feelings of confusion, despair and uncertainty, which appear to intensify the longer the pre-diagnostic period lasts (36). The appearance of first symptoms and the subsequent quest for a diagnosis disrupt the structures of everyday life and it's taken-for-granted features. Diagnostic delays are common in families' narratives, augmenting feelings of insecurity, and uncertainty, where frequent medical controls, extensive hospitalizations, and long-distance traveling to access specialized centers makes normal family life impossible (8).

The difficulty in obtaining the correct diagnosis is the first dramatic hurdle for rare disease patients and may take years, or even decades to overcome. When seeking diagnosis, patients frequently consult numerous doctors, undergo multiple examinations, and often receive various incorrect diagnoses resulting in inefficient and even harmful treatments (19).

The parental experience of searching for a diagnosis has been described as a journey with two distinct components (41):

- An inner emotional experience that includes the realization that there is a problem, wanting a diagnosis and coping with it
- An outer sociological experience which includes experiences with professionals and support networks

This diagnostic odyssey can be a significant source of parental stress and uncertainty, the psychosocial effects of which remain poorly characterized (41). Consequences of delayed diagnosis include anxiety, loss of reproductive confidence because of an ill-defined genetic risk, frustration and stress, disease progression, delays in treatment and inappropriate treatments (3).

From the point of view of professionals and parents, the perceived reasons for diagnostic delays included lack of knowledge about the disease among health professionals, lack of symptom awareness by the family, and difficulties accessing tests (3). Parents often believe their child's diagnosis could have been made earlier, citing reasons such as lack of knowledge by health professionals about the disease, and unavailable or delayed testing (1).

A definitive diagnosis of a rare disease is a double-edged sword. The experience of the diagnosis announcement was reported by parents as an emotional "shock", a "relief" or "both a shock and a relief", regardless of the possible inheritance (7). Advantages of obtaining a conclusive diagnosis included becoming more accepting towards the situation, being enabled to attune care to the needs of the child, and better coping with feelings of guilt. Disadvantages experienced included a loss of hope for recovery, and a loss by parents of their social network of peers around the time of diagnosis, and the effort necessary to re-establish that social network (21).

Biomedical diagnostic approaches (such as Whole Exome Sequencing [WES]) are not a panacea (21). They offer a biomedical description to parents, explaining the genetic cause for malfunction or disease, which some parents use to deal with the social dimension of having a diseased child; a label for the condition of their child allows parents to explain the child's behaviour to the outside world, organize adequate care, and identify with a supportive group of peers (21). The lack of information about each specific rare disease often means that the definitive ending of the diagnostic odyssey by WES opens up another odyssey in the search for information, new care arrangements, and search for new peer contacts. This is often accompanied by the loss, and associated mourning, of previous contacts and feelings of guilt and worry associated with knowledge of the mutation (21). It is essential that genetic counsellors prepare parents for these difficulties before the test and continue to support them after test results have been obtained (21).

Poor Diagnostic Delivery

The majority of families started their lived experience from the time they received their child's diagnosis; this diagnosis, and the manner in which the diagnosis was given, then impacted their life as a whole (33). The *'tone'* of how families experience their journey with lived experience is often set at diagnosis, and heavily influenced by the manner of its delivery. Some families may be dissatisfied with the way the diagnosis is delivered, citing lack of empathy and information from health professionals (1, 3, 23, 32).

The diagnosis is seen as a turning point that *could* enable families to start adjusting to their new normality (8). Unfortunately this experience is often characterised by some, or all of being: left

alone, with no offers of additional counselling or support, in a room crowded with professionals, and those professionals leaving mid-way through the diagnosis, that is delivered quickly, often accompanied by an equally off-hand referral to another clinic (1).

The manner in which the diagnosis announcement is made plays an important role in the way people react (42), it is common that the manner causes the parents to perceive that the healthcare professional simply does not appreciate how devastating receiving a diagnosis of a rare and complex disease can be (3), which instantly undermines trust in any information then imparted.

It is not too strong to describe the manner of delivery of the diagnosis as *'traumatic'* for the recipients, and many show signs that are akin to PTSD. With families forced to repeat experience of loss, feelings of pre-bereavement loss and chronic sorrow, in subsequent encounters with health services that are especially linked to tone of delivery of diagnosis.

Lack of Information Provision

Experiences immediately after diagnosis were described as overwhelming, scary, and lonely, with little information or support from health care providers (17). Parents living with a child with a rare, presumably genetic disease are often confronted with uncertainties because they lack good-quality information about the condition, prognosis, therapy, and recurrence risk of the disease (21). Families can be left to their own initiative to find information without guidance or come across it by chance.

Parents of children with a rare disease voice frustration at the lack of knowledge and experience demonstrated by healthcare professionals associated with their child's disease (9). The scarcity of information provided by healthcare professionals leads to frustration and disappointment, particularly with the lack of individualised information and support offered to their child, themselves, and their families (23). This scarcity of information leads to feelings of isolation and uncertainty, compounded by the apparently extensive information that is readily available and provided for other childhood illnesses (23).

Isolation is predominant in the rare disease community, so many parents come to view connections to an online community for support as critical, as a route to access information, increase knowledge and empowerment, advocate for their child, and establish relationships with others going through a similar experience, especially as this is not readily forthcoming from health services (17). Parents' support groups fulfil a precious function of peer support, and an information source for parents of children with rare diseases (39). However, the information from unmanaged sources including social media often triggers chronic sorrow through negative information and information overload (17).

Poor communication by healthcare professionals, and their inability to provide parents with a comprehensive prognosis for them and their child are a constant source of concern for parents (9).

Parents will often feel a sense of loss for the life they were used to before the disease and may yearn for what they perceive as a normal family life (8), and parents feel that a lack of information from healthcare professionals directly hindered their ability to cope and manage their child's health when situations arose (9).

Lack of Specialist Knowledge

Healthcare professionals' limited knowledge of the obstacles faced by rare disease patients, and healthcare structures' limited capacity to accommodate rare disease patients, who have needs that differ from others, leads to actions by healthcare professionals that are perceived as rejection (19).

Lack of healthcare professionals' knowledge of the disease affecting their child remains a commonly cited cause, by parents, of late diagnosis (1). This lack of knowledge creates a vicious circle when combined with testing. Tests are unavailable so healthcare professionals do not become aware of the diagnosis; because they are unaware, they do not make tests available. Even when tests are available, without knowledge testing is often overlooked or delayed (1).

There have been repeated calls for better education of health professionals during undergraduate and postgraduate studies, better awareness of existing information resources and development of new resources to support clinical care, and resources including clinical guidelines on diagnosis and treatment, lists of specialised clinics and referral pathways (1). Increased knowledge and understanding of the impact of living with a rare disease is needed in order to improve good practice guidelines for the care and support of families and professionals (12).

Lack of specialist knowledge leads to a lack of information provided to patients and their families at the initial diagnosis, leaving them to do their own. The knowledge and information required is different to that required by parents of children without a rare and complex disease, and often extends to specialist health knowledge and additional health literacy, care giving skills, and resources beyond those normally required by parents to effectively parent their child (25).

Unassessed Psychological Need

Not all parents of children with disabilities have difficulty adapting, however, elevated levels of stress almost become normal in the parent's life. Psychological support seemed to be variably proposed, and needs are only partly evaluated or taken into consideration (42). Parents caring for children with rare diseases fear the long-term progression of their child's disease (13).

The psychosocial impact of a diagnosis is moderate to high for nearly all (90%) families, and the level of impact is not dependent on the level of health functioning of the child (1). Families caring for children with genetic metabolic disorders are particularly adversely impacted by delays in diagnosis, lack of easy access to peer support groups and a lack of psychological support (1).

Few families are offered psychological support or counselling at the time of diagnosis (3), despite feeling 'devastated', 'confused', 'heart-broken' and 'in shock' when given the diagnosis (1). The vast majority believe that psychological support should always be provided (3); yet even when psychological support is offered, it is rarely given by specialists (3).

Maze-like Healthcare Systems

Bureaucratic problems are frequently encountered when navigating social and health care systems (8). Families feel particularly vulnerable due to the fact that collaboration and communication between healthcare service providers are often fragmented and unsatisfactory (33). Healthcare systems evoke feelings of disbelief and frustration, as they show little understanding of what challenges parents of children with rare and complex diseases experience in their day-to-day life (33).

Parents describe the current healthcare system as being like a revolving door, in which they repeatedly enter and exit, each time having to tell their story to a different professional (33). Currently, healthcare professionals with expertise in a given field will provide care for specific elements of the condition, rather than being concerned with the condition as a whole (33). This can lead to bureaucratic complications when attempting to navigate the social and healthcare systems and to feelings of vulnerability, being lost within the system and falling through the net (8, 33).

Ignored Practical Needs

The severity of a child's condition is associated with work-related changes and changes to a couple's relationship, with increasing severity associated with more negative consequences of work-related changes and economic changes, than with other life changes (31). These impacts mean that the severity of the child's disease is negatively associated with parental Health-Related Quality of Life (HRQoL) (37). Parents often must take a leave of absence, or even sacrifice their careers to learn and adapt to the complexities of the disease (30), as these practical needs are (or appear to be) ignored by healthcare professionals. Consequently, social support, including from close family and friend relationships, and social media, are important to caregiver and family well-being (30).

The costs of caring for a child with a rare disease can be significant. In addition to day-to-day care, families may need to pay for travel costs, and adaptations to the family home to cater for their child's wheelchair needs (21). Some parents reported using the internet and social media to access resources, and to connect with other families going through similar hardships. Support from family, friends and peers was considered vital for family wellbeing (30).

Forgotten Family Members

The psychological health of the entire family is impacted by having a child with a rare disease (13). Mothers of affected children are more likely to suffer with poor mental health and impaired quality of life compared to fathers (13). Indeed, the parents of an affected child have to accept that their own desires may not be met or achieved, and they may experience a lower level of life satisfaction (19). The mental and emotional toll on parents is also commonly shared by siblings, who experience a complex range of contradictory feelings including guilt, pride, worry, and sadness (19). The daily lives of siblings of children with a rare disease can be greatly impacted if family life is centred around caring for their brother or sister, and attention must be paid to support their emotional and social needs (19).

MAKING A POSITIVE IMPACT AROUND THE TIME OF DIAGNOSIS

Parents of a disabled child will have to achieve a range of things: managing their child's afflicted health, accepting the child's condition, managing the child's condition on a day-to-day basis, meeting the child's normal developmental needs, and meeting the developmental needs of other family members (15). However, it is a rare person who appreciates this at the time of the diagnosis, and most will at some point require some form of support whether in accepting (16),or meeting these needs (21).

Most families want help seeking knowledge about a child's condition and prognosis, caregiving activities and management of care, and finding a supportive environment (16, 21). Their needs might be grouped in to three main themes: the need for normality and certainty, the need for information, and the need for partnership (16, 43).

Interventions for parents can both change factors that contribute to parental stress, perceived strain, and the sources of life satisfaction (15), as well as lessen stress, perceived strain, and improve life satisfaction. Professional teams are hugely important to develop parents' knowledge and skills concerning their child's disability (15). Increased parental knowledge and more active coping leads to better use of resources in the family, social network, and wider society (15).

A Definitive Diagnosis

A definitive diagnosis is often difficult (9), however, a conclusive diagnosis allows for parents to progress in their lives. Parents with conclusive diagnoses are able to re-establish a peer community (21). Parents describe an emotional rollercoaster of fluctuating emotions that alternate between a happy confidence of being in control and a sense of anxious uncertainty; this sense of control is associated with a confirmed diagnosis and agreement on treatment plans (23).

Sensitive Sharing of the Diagnosis

Conditions under which the rare disease diagnosis is made is as important to patients and their families as the announcement of the life-changing diagnosis itself, and is crucial in helping patients and their families accept the diagnosis and commit to appropriate treatment (19).

The way that parents cope and adjust to their child's diagnosis is pivotal, not only to the family's physical and emotional well-being, but also to the child's own coping and adjustment to the disease (8).

Parents need support from healthcare professionals to help them adapt to the multisystemic problems caused by their disease, in coping with its challenges (40), and to ameliorate the quality of life impacts of the multiple demands of caring for a child with a rare and complex disease (20). This support needs to include support physically, socially, emotionally and psychologically (40).

Time, information and compassion are the basis of a positive experience (1). Ideal conditions to sensitively impart diagnosis of a rare disease include comfortable conditions for patients and their families, along with adequate time (to explain the nature of the disease, its consequences on daily life, the disease progression and prognosis, as well as adequate time for patients and their family members to freely ask any additional questions) (19). A simple basis for meeting these needs could begin with a consultation specifically dedicated to the delivery of the diagnosis (19).

Provision of Managed Information

A lack of information given to parents, often leads to them going out of their way to do their own research, this can be seen as a coping technique, to combat uncertainty trying to gather as much information as possible; for example, by visiting one medical specialist after another, by searching the internet, and also by exchanging information with other parents with children with similar complaints via social media (21). However, this coping mechanism can easily slip and become pathological; this slipping is best prevented through the pro-active sharing of high-quality, curated information and explanation. Most parents would like information on a website (16).

Families highlight the need for more knowledge about taking care of a child with possible special health care needs, with information that is consistent and clear (22). The importance of involving patient organisations and their representatives in the construction and delivery of the information cannot be underestimated (19), this is particularly true in where specialist knowledge, time, or support are limited in health systems. Increased parental knowledge and more active coping leads to better use of resources in the family, social network, and wider society (15). Increasing education of parents increases their efficacy for coping with stress (11).

Reliable information is scarce and/or difficult to find (8) so healthcare professionals must actively bridge information to parents, especially as they easily develop a fear of asking questions as they feel that they are perceived as annoying (23). As healthcare professionals share knowledge, care must be taken to tailor it to impart understanding, rather than simply information, as freely used medical terminology can be incomprehensible for parents (7).

The need for managed (curated) information is not limited to patients. There is also a need for better education for health professionals (including specialists), especially resources to help with clinical care, such as clinical guidelines on diagnosis and treatment, educational modules, and referral pathways (1).

Professional Support

While parents may express a preference for face-to-face support, a combination of face-to-face and online sessions is also considered appropriate. Most parents would prefer to have at least the first session face-to-face, followed by online sessions. It is important that interventions are guided by professionals (16). Group interventions allow for sharing of experiences and tips with other parents

in a similar situation (16), thus augmenting the support of professionals, with opportunities to build wider support networks.

Even when parents do receive a lot of clinical information when their child is diagnosed, they experience trouble finding their way into psychosocial support and/or contact with other parents (16). A standard consultation with a psychosocial healthcare provider (e.g. psychologist, social worker) a few months after the child's diagnosis would be welcomed by parents, and all interventions, should be easily accessible (16).

Examples of good practice include the use of Narrative Intervention post diagnosis check-ins by trained nurses/professionals (18).

Guidance of Health System Navigators

Parents in families affected by rare diseases have a near ubiquitous experience of problematic navigation of health care systems (8). Most health systems leave people to attempt this navigation alone, and few have formal resources in place to facilitate smooth navigation. When asked, parents said that they would like their General Practitioner to coordinate services. Co-ordinating services requires knowledge of the condition, and whilst currently most General Practitioners do not have sufficient knowledge to perform this role, parents agreed that their General Practitioner could have enough knowledge on their child's disease (1).

Teaching Coping Techniques

There are many ways of coping with stress: physical techniques such as exercise and rest, cognitive techniques using faith or a belief to help cope, or behavioural techniques using activities to help decrease stress (11). Coping can be more broadly thought of as adaptive/constructive behaviours, typically classified into three types: appraisal focused, problem focused, and emotion focused (17).

More psychosocial support is needed as parents adjust to a new diagnosis (18). Some parents are able to cope effectively and become strengthened by the challenge, while for others, the challenge exceeds the family's resources (15). By supporting parents well in this first intensive period, parents will likely become able to support their own child (14) .

To manage the burden of care, parents' needs are summarized in three main themes: the need for normality and certainty, the need for information, and the need for partnership (15). The process of normalization is a part of coping, and when stress is effectively managed, deeper coping becomes possible (8).

Interventions have been designed and tested in some conditions (such as childhood Type 1 diabetes (18)) with stepped support, including social support, brief phone-delivered cognitive and behavioural support, and individualized consultation, according to need.

Training and information is important to facilitate coping (26) as the provision of information fosters problem-focused coping (26). There is an important role for physicians, who often need training to deliver this role, in helping parents develop appropriate strategies for 'seeking support' (26).

Some intervention programs (based on Cognitive Behavioural Therapy) are available that involve parents. However, those interventions are often primarily focused on teaching the parents to support their child managing the illness (16).

Studies have shown that sharing experiences with others in a similar situation is associated with a decrease in distress, and an improvement of mental health for children and parents (16). There are great benefits seen for parents who are able to use focused stress coping strategies and start moving away from emotion to problem solve (11, 12).

Specific Psychological and Psycho-Social Interventions

There is a need for routine psychological support following diagnosis (1). The focus of psychological and psychosocial interventions is more effective when it is on parents themselves, as opposed to many existing parental interventions that focus on teaching parents how to support their children (16).

Individual face-to-face counselling with a therapist in the hospital is attractive to many parents. Group interventions where they can share experiences and tips with other parents in a similar situation are also favoured. Parents prefer face-to-face support; however, a combination of face-to-face and online sessions would also be appropriate. Most parents would prefer to have the first session face-to-face, followed by online sessions. It is important that any interventions are guided by professionals (16).

Parents have trouble finding their way into psychosocial support and/or contact with other parents. Moreover, they find it hard to seek and accept psychosocial support; considering consulting a psychosocial healthcare specialist as a failure and feeling like they have to solve the problems themselves. Parents would like a standard consultation with a psychosocial healthcare provider (e.g. psychologist, social worker) a few months after the child's diagnosis and have emphasized the need for intervention to be easily accessible (16).

It is important that counsellors are aware that even if a diagnosis is long sought and longed for, it does not bring prolonged relief, as the relief quickly transitions into feelings of uncertainty and loss (21).

Psychosocial support services should aim at strengthening the family situation, overcoming social isolation, strengthening intra-familial relationships, and developing coping strategies for the handling of the situations emerging from the child's disease (13).

Further Considerations

During the development of this work, other important factors became apparent that fell out of the scope of the selected literature. These warranted further investigation and are summarised in this section.

Framework for sharing bad news

The delivery of a diagnosis should use some form of validated framework to support the process. An example of this kind of good practice is the SPIKES framework for delivery of news (**S**etting, **P**erception, **I**nvitation and **I**nformation, **K**nowledge, **E**mpathy, and **S**ummarize and **S**trategize) (44). Delivering news is a complex communication task, following the SPIKES framework can help ease the distress felt by the patient who is receiving the news, and the healthcare professional who is delivering it. Key components of the SPIKES strategy include demonstrating empathy, acknowledging and validating the patient's feelings, exploring the patient's understanding and acceptance of the bad news, and providing information about possible interventions. Having a plan of action provides structure for this difficult discussion and helps support all involved (44).

Assessing psychosocial risk factors

The importance of psychological support is highlighted by the fact that many parents of children with a rare and complex disease score above the cut off on a screening instrument for depression (5). The psychosocial profile of caregivers is characterized by high levels of burden and burnout (45, 46), parental stress and positive adaptation processes despite the loss of health (45), deterioration in family functioning (47), symptoms of depression (45), symptoms of anxiety (48), negative coping styles (49), low levels of resilience (50), little social support (51), optimism (52), and effects on quality of life (34). Family cohesion also provides an indirect indication of an important additional dimension (53).

The vulnerability of family caregivers is characterized by caregivers with low levels of family support, resilience and well-being, and high levels of anxiety, overload, depression, and parental stress. This profile identifies care-givers at risk of presenting psychopathologies that affect the quality of the care they provide to the paediatric patient (34).

Social support

The experience of rare and complex disease is daunting (54), and social and psychological support is essential (55). Caregivers experience feelings of guilt when delegating caregiving to other family members and/or professionals (12). Social support can help alleviate depression, increase sense of self-esteem, increase coping strategies, and elevate life satisfaction and psychological well-being of an individual (4); with effective social support networks positively influencing the quality of life of caregivers (56). Coping with stress and traumatic events is positively influenced and

moderated through the social support individuals receive or perceive they have access to (36, 57, 58). There is additionally a specific need for someone families can approach with questions regarding the condition they are living with (59).

It is not uncommon for patients to have never met someone with the same condition until they come across a dedicated support group (59). Patient organisations can often be the only accessible source of up-to-date reliable information on the condition and are uniquely placed to offer support to newly diagnosed patients. Patient organisations can be a huge source of information for patients and families, so it is vital that people are made aware of them as soon as possible (59). Self-help organizations fill a gap and complement the work of physicians by informing patients, guiding them through the health system and offering a partner to talk to (2). Parents and children demonstrate improved adjustment and increased social support when a parent mentor is available, and there is evidence of better health outcomes where these mentors are part of families' support systems (18).

Online communication provides the critical mass of people needed for support and facilitates finding information about rare conditions (17). Caregivers enjoy being part of an online community and meeting others in similar circumstances, particularly in support groups and social media, but online communities cannot completely replace in-person interactions (17).

A key set of opportunities for building support networks are contained in family and scientific conferences. Patient organisations are often the most effective group to organise these. Repeated conference attendance is correlated with higher companionship, emotional, and informational support (60), and in the field of rare diseases is associated with improved self-efficacy and an increase in perceived knowledge (60). Specifically, being surrounded by others who share experiences with the impact of one's condition offers a unique opportunity for destigmatizing companionship support, which normalizes a person's experience, reduces isolation, and promotes solidarity (60). Hundreds of millions of people worldwide with rare diseases face unique challenges to quality of life (QoL), including stigma and limited support. To address these concerns, many rare disease organizations offer support conferences for people to meet others with their condition (55).

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ACKNOWLEDGEMENTS

Rare Disease Research Partners would like to thank the expert group for their invaluable insight and assistance during the development of this work

This work was supported in full by a grant from RBW Consulting - a life sciences talent & strategic consultancy. For more information about RBW Consulting please visit: www.rbwconsulting.com

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Date of preparation: November 2021