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Rare Disease Research Partners

Psychological Support for Parents and Caregivers of Children with a Rare Disease Diagnosis - Consensus Statement of Good Practice

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CONTENTS

ABSTRACT

The diagnosis of a rare disease has a significant psychological impact upon the family and caregivers of the affected child. Upon receiving a definitive diagnosis, parents often feel initial relief, which is soon replaced by overwhelming feelings of stress and guilt. Stress often manifests as anxiety, uncertainty, worry, fear, frustration, grief, powerlessness, shock, or denial. The purpose of psychological support is to reduce the mental and emotional burden that parents and caregivers experience when confronted with a diagnosis of a rare disease. Currently, there are no published consensus guidelines on the type and extent of psychological support needed by families and caregivers of children affected by a rare disease at the time of diagnosis. A comprehensive literature review was undertaken, and this statement was developed by experts from the UK and USA including clinical and mental health experts and parents of children with a rare disease. Recommendations were put forward for the delivery of a diagnosis, information provision, access to disease expertise and psychosocial support, facilitation of support networks, and co-ordination of care.

INTRODUCTION

Rare diseases affect approximately 6% of the population and are notoriously difficult to diagnose and treat [1]. Patients frequently experience diagnostic odyssey; a long and convoluted path to a definitive diagnosis. Families often wait years from the onset of first symptoms to diagnosis, causing significant delays in treatment, unnecessary tests, and psychological stress for the family [1,2]. Recent studies have provided valuable insights into patients living with rare and undiagnosed conditions, including the effect on their mental health [3,4]. Another newly published study explored the emotional impact of the diagnostic process on adult patients and their families [5]. However, the information and guidance available for parents and caregivers of children with rare diseases specifically around the time of diagnosis remains scarce.

The impact of a rare disease diagnosis is psychologically complex, parents are often torn between feelings of relief, and feelings of sorrow, loss and/or guilt [6]. Families regularly reported initial relief at reaching a diagnosis, and these feelings are often intensified in those families that have endured a long diagnostic delay [7]. Parents and carers typically described the period surrounding diagnosis as a highly emotional time for them. Parents often feel overwhelmed with sadness, feelings of vulnerability and suffered from anticipatory loss, blame, confusion, disbelief, dismay, helplessness, insecurity, and perceive a lack of control [8].

Following diagnosis, there appears to be a substantial lack of knowledge to support those affected [9], and families and caregivers often take on the role under sudden and extreme circumstances, with minimal preparation and little guidance and support from healthcare systems [10]. Parents of a disabled child will have to cope with managing their child's condition on a day-to-day basis, whilst

meeting their child's normal developmental needs in addition to the developmental needs of other members of the family [11]. There is a risk that the caregiver burden may become too great, and their well-being compromised, meaning they will no longer be able to provide the vital care required by the child [10]. Caregivers experience high levels of stress, depression, and anxiety [12], and there is a significant psychological impact on them, which in turn affects other members of the family [13]. 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].

Here, we propose a set of recommendations for the psychological support provisions necessary to improve longer-term outcomes by providing crucial psychological support at the time of diagnosis.

SCOPE OF THE STATEMENT

This statement is intended to address the unmet need for psychological support, by raising awareness and providing guidance for healthcare and other supporting professionals on the extent and nature of intervention and provision needed by parent and caregivers at the time of diagnosis. The scope is diagnoses of rare diseases made in infancy and early childhood, although the findings and their implications are likely to be more widely applicable.

METHODS

This statement is based on evidence generated from a review of the literature conducted systematically and the consensus of a group of experts. The literature review yielded 37 relevant published articles [1,2,7,8,11,14-45]. The expert group comprised of healthcare professionals, patient representatives, and those directly affected by a rare disease.

Evidence to support the recommendations has been graded using the following definitions:

- Strong evidence: consistent evidence and new evidence unlikely to change recommendation and expert consensus
- Moderate evidence: expert consensus or majority decision but with inconsistent evidence or significant new evidence expected
- Weak evidence: inconsistent evidence AND limited expert agreement

RECOMMENDATIONS

The literature reviewed gave a clear and stark picture of the potential for adverse psychological and psychosocial impacts during the time of, and through the process of receiving a diagnosis for a rare and complex disease. How to correct this, however, appears to be less researched. Many examples of good practice have not yet made it into the published literature, so in addition to the explicit recommendations, it would be enormously beneficial to see the communities of researchers and practitioners share additional evidence of beneficial intervention around the time of diagnosis.

It is also apparent that there are additional and specific needs for families as they journey and live without a diagnosis. There would be significant value in similar future investigations of the literature that draw together that evidence. There are additional and specific needs for families as they live following a diagnosis, and there are many rare and complex diseases for which there are unique adverse psychological and psychosocial impacts and associated needs. There would be great value in future research and specific examination of the literature to illuminate and develop evidence to enhance and guide support, care, and targeted interventions. The following practical recommendations are derived from the current published evidence.

Delivery of the diagnosis

There was often a long delay between the onset of first symptoms and diagnosis. Many parents described the period from the appearance of first symptoms to diagnosis as the most difficult and psychologically painful [7,41]. By the time of diagnosis, families affected by a rare and complex disease were best characterised as emotionally and financially stressed, with costs associated with caring for their child [30].

The tone in which the diagnosis was delivered to the family had a heavy influence on their prospective journey. Some families have reported traumatic experiences at diagnosis, citing rushed delivery, lack of empathy, being left alone, or being left in a crowded room of professionals and of those professionals leaving mid-way through the diagnosis [1,2,27,37].

Delivery of the diagnosis

Recommendations	*Grading*
1. Diagnosis should be delivered by a health professional with substantial knowledge of the condition and with training in how to deliver difficult news	Strong
2. The healthcare professional should allow sufficient uninterrupted time to deliver the diagnosis in a sensitive and unhurried manner, allowing for the information to be processed	Strong
3. When more than one professional is involved in delivering the diagnosis, all professionals should remain with the family throughout the entire consultation	Moderate
4. A private, comfortable, quiet space with room for the family, and all the healthcare professionals involved to remain during the diagnosis should be provided	Moderate

A reliable source of curated information

Parents described feeling overwhelmed immediately after diagnosis, with little information about the condition, prognosis, and therapy from health care providers [19,25]. Accurate information was difficult to find without guidance and families were often left to research this for themselves [46]. Information from unmanaged sources including social media often triggers chronic sorrow through negative information and information overload [19].

A reliable source of curated information

Recommendations	*Grading*
1. Parents and caregivers should have access to a source of accurate information that is frequently and regularly reviewed and updated so it remains relevant and useful	Strong
2. Signposting for parents and caregivers to relevant and accessible financial and mental health support services	Weak

Access to expertise

Reliable information is scarce and/or difficult to find [7] so healthcare professionals must actively bridge information to parents [27]. As healthcare professionals share knowledge, care must be taken to tailor it to impart understanding, rather than simply information [22]. Parents felt that a lack of information from healthcare professionals directly hindered their ability to cope and manage their child's health when situations arose [8]. The need for managed (curated) information is not limited to patients. There is also a need for better education for health professionals [1].

Access to expertise	
Recommendations	*Grading*
1. No single source alone can support the information needs of families affected by rare and complex diseases. Information needs contextualisation and interpretation to become useful knowledge. This interpretation and contextualisation are best done by people with substantial experience, knowledge and expertise surrounding the care, impact and prognosis of a given disease	Strong

Access to a psychological healthcare practitioner

The psychosocial impact of a diagnosis was moderate to high for nearly all (90%) families [1]. Families were impacted by delay in diagnosis, lack of easy access to peer support groups and an absence of psychological support [1]. Few families were offered psychological support or counselling at the time of diagnosis [2], 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; yet even when psychological support is offered, it is rarely provided by specialists [2].

Access to a psychosocial healthcare practitioner	
Recommendations	*Grading*
1. Psychological assessment and support delivered by a trained, experienced healthcare practitioner should be readily available to all families from the time of diagnosis onwards	Strong

A single (or named) professional with clear responsibility for co-ordination of care

Families often encountered fragmented communication between healthcare providers, with incidents such as repeated loss of medical notes in clinics [46]. These difficulties with navigating the social and healthcare systems lead to feelings of disbelief, frustration, and exhaustion as they show little understanding of what challenges parents of children with rare and complex diseases experience in their day-to-day life [38]. Often families find that each professional looks at specific elements rather than the condition as a whole, leading to families having to repeatedly tell their story to each professional involved in their care [41,46].

A single (or named) professional with clear responsibility for co-ordination of care	
Recommendations	
1. Most health systems have a professional (e.g., General Practitioner) who implicitly or explicitly co-ordinates care. In rare and complex disease, that professional needs additional support to gain sufficient disease and service knowledge to effectively co-ordinate care	Moderate
2. Experts need to actively share disease knowledge with the co-ordinating professional	Strong

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Rare Disease Research Partners
MPS House, Repton Place, White Lion Road
Amersham, Buckinghamshire, HP7 9LP, UK
t: +44 (0) 345 260 1087
e: info@rd-rp.com

w: rd-rp.com

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