Review

The Importance of Psychological Support for Parents and Caregivers of Children with a Rare Disease at Diagnosis

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SUPPLEMENTARY MATERIAL

Supplementary Table 1. Summary of selected studies.

Study	Disease/disorder	Study Participants	Key Findings
Anderson *et al*., 2013 (1)	LSD; mitochondrial disease	46 families	Over 35% of patients saw 3-5 doctors before receiving the correct diagnosis, and 43% of patients thought the diagnosis was delayed. Some families were upset with the delivery of the diagnosis, reporting insensitive communication style, inadequate information given and a lack of psychological support. Approximately 90% of families cited moderate to high psychological impact. Twenty-six (87%) wanted, but only 13 (43%) received, information about peer-support groups.
Atasoy & Sevim, 2018 (11)	Mental disability	302 parents	Parents were better able to cope with stress by increasing their understanding of the disorder. Emotion-focussed coping strategies were commonly used.
Berrocoso *et al*., 2020 (12)	Wolf-Hirschhorn syndrome	22 caregivers	Negative impact on QOL was associated with symptomology. The use of engagement strategies such as problem-focused coping was associated with improved psychological QOL and social support.
Boettcher *et al*., 2020 (13)	Rare diseases requiring long-term ventilation	75 families	Mothers' QOL and mental health were severely impacted. Protective factors include coping mechanisms, social support, and family functioning. Multiple regression analyses showed that family functioning might be the most important predictor of QOL and mental health.
Dellenmark-blom *et al*., 2016 (14)	Oesophageal atresia	30 families	Nine coping strategies were identified: problem solving, avoidance, recognising responsibility, confronting, seeking social support, positive reappraisal, emotional expression, acceptance, and distancing.
Dellve *et al*., 2006 (15)	Rare diseases	246 parents	High parental stress, physical and emotional strain was found among mothers, especially among single mothers. Fathers showed high stress, related to incompetence. An intensive family competence intervention improved some aspects of parental life satisfaction.
Douma *et al*., 2020 (16)	Children with chronic illness	272 parents	The aim of the study was to refine an existing face-to-face intervention into an online psychosocial group intervention. The study found that parents preferred face-to-face contact, but after an explanation and demonstration of an online intervention, parents became more positive about online support, mostly because they could participate from home.
Germeni *et al*., 2018 (6)	Bartter syndrome; celiac disease	26 mothers	Most participants experienced relief when given a definitive diagnosis, and comfort from access to appropriate information, social support and contact with other families.

Glenn 2015 (17)	Alagille syndrome	16 mothers	Findings suggest mothers need emotional support and help to access appropriate online resources.
Heuyer *et al*., 2017 (41)	Rare diseases	448 respondents	Large disparities in the delays to obtaining a diagnosis were reported (1 year to >20 years). Professional lives of patients and parents were strongly and negatively impacted by the disease. Psychological issues were reported by 62% of respondents.
Hilliard *et al*., 2017 (18)	Diabetes Type 1	Caregivers of children with Diabetes Type 1	Paper reports on the development and study design of First STEPS: Study of Type 1 in Early childhood and Parenting Support.
Houdayer *et al*., 2013 (7)	pCNVs e.g. Williams Syndrome	60 parents	This study aims to describe the psychological impact of the diagnosis announcement of pCNVs. The announcement of inherited pCNVs can increase the feeling of parental guilt. The study shows that the disclosure of pCNVs has a significant psychological impact.
Kole & Faurisson 2009 (2)	Rare diseases	12, 000 patients	This study reported that 40% of patients were initially misdiagnosed, leading to severe consequences such as inappropriate medical interventions, including surgery and psychological treatment. One-quarter of patients reported waiting between 5-30 years from the appearance of the first symptoms to receiving the correct diagnosis. In almost half of the cases, the diagnosis was reported as being announced under inadequate or totally unacceptable conditions. The study found that a general lack of awareness and a specific lack of knowledge of rare diseases in healthcare professionals may be the result of inadequate training during medical studies, reflecting a need for strengthened advocacy and continued education.
Kountz-Edwards *et al*., 2017 (19)	Juvenile dermatomyositis	36 mothers; 3 fathers	Families reported difficulties in family functioning, communication, and an increased number of conflicts. Parents appeared to be experiencing higher than average levels of worry, worse physical functioning, and family relationships.
Krabbenborg *et al*., 2016 (20)	Kabuki syndrome; MECP2 duplication syndrome; 16p microdeletion syndrome; congenital Rett syndrome	Parents of 15 children	This study found that the advantages of obtaining a definitive diagnosis included becoming more accepting of the situation, being able to attune care to the needs of the child and coping better with feelings of guilt. Disadvantages experienced included a loss of hope for recovery, and a loss by parents of their social network of peers.
Lakshmanan *et al*., 2019 (21)	Infants with medical complexities	21 participants	Mothers felt great anxiety and stress when comparing their situation to those of their peers. Families often found hope and resilience in peer support.
Lauder *et al*., 2018 (22)	Early onset scoliosis	12 mothers	Four major themes emerged: emotional rollercoaster ride, a lack of resources, money talks and pervasive burden. The findings suggest there are multiple factors that influence the experience of mothers' caring for a child with early onset scoliosis.
Merker et al., 2020 (23)	Schwannomatosis	18 patients	The study identified three elements of effective diagnostic communication: education (aetiology, prognosis, and treatment options); psychological support (to cope with the new diagnosis and any prior diagnostic harms); and efforts to develop therapeutic

			alliance (feelings of collaboration, trust, and social-emotional rapport). Poor communication was characterised by inadequate or jargon-heavy explanations, perceived disinterest in or disbelief of symptoms. Effective communication helped people feel informed and cope with their condition; poor communication caused significant psychological distress.
McConkie-Rosell *et al*., 2018 (40)	Undiagnosed diseases	50 parents	Study found that parents of undiagnosed children had high rates of anxiety and depression (~40%), which were negatively related to coping self-efficacy.
Pelentsov *et al*., 2016 (24)	Rare diseases (132 distinct rare diseases)	301 parents	Study found that 54% of parents were dissatisfied with health professionals' level of knowledge and awareness of disease; 71% of parents felt they received less support compared to other parents. Information regarding present (60%) and future services (72%) available for their child was considered important. Almost half of the parents struggled financially, 38% reduced their working hours, and 34% ceased paid employment. Forty-two percent of parents had no access to a disease-specific support group, and over half stated that their number of friends had reduced since the birth of their child; 75% had no contact with other parents with a child with a similar disease, and 46% reported feeling socially isolated and desperately lonely. Most frequent emotions expressed by parents in the week prior to completing the survey were anxiety and fear, anger and frustration and uncertainty.
Pelentsov *et al*., 2015 (8)	Rare diseases	Review of 29 studies	Most common needs of parents caring for a child with a rare disease were social needs (72% of papers), followed by informational needs (65% of papers) and emotional needs (62% of papers). The most common parental needs overall were information about their child's disease, emotional stress, guilt and uncertainty about their child's future health care needs, parents' own caring responsibilities and the need for more general support.
Picci *et al*., 2013 (25)	Rare diseases and chronic diseases	111 parents	Parents of children with chronic diseases preferentially used problem-focused and emotion-focused strategies (active coping, seeking instrumental social support, and positive reinterpretation and growth), whereas parents of rare diseases often turned to religious coping. The uncertainty surrounding rare diseases appeared to impede the use of active coping.
Rice *et al*., 2020 (26)	Rare diseases	Review of 34 studies	All included studies reported on the benefits of participating in psychosocial interventions. Benefits that were most commonly found included statistically significant improvements in emotional states (e.g. stress) and caregiver burden and narrative reports of intervention helpfulness. Statistically significant improvements in mental health outcomes (e.g. depression symptoms) were rarely detected.
Senger *et al*., 2016 (27)	Mitochondrial disease	231 mothers	On average, children had seen seven different specialists; 61% were hospitalised in the past year. Significant correlations ($p<0.05$) were found between parenting stress and parent age, parent income, parent education, child age, child age at diagnosis, presence of developmental delays, number of hospitalisations, number of medical visits, number of organs involved, and number of specialists seen. Significant correlations were also

			found between parenting stress and coping behaviours such as family integration, social support and understanding of health care.
Shapiro *et al*., 2019 (28)	Sanfilippo syndrome type B	International clinical advisors meeting	Important factors contributing to caregiver burden included sleep disturbances, impulsive and hyperactive behaviour, and communication difficulties. Caregiver burden remained high throughout the life of the patient, and coupled with the physical burden of daily care, had a cumulative impact that generated significant psychological stress.
Siddiq *et al*., 2016 (29)	Inherited metabolic diseases	21 parents	Most parents reported that they and their families had adapted well to their child's diagnosis. Proactive coping strategies were used to integrate complex disease management protocols into routine family life. Social challenges faced by their children were a source of parental stress. Participants reported positive interactions with most healthcare providers but reported challenges with the healthcare system outside of disease-specific metabolic care, when encountering providers unfamiliar with the child's disease.
Silibello *et al*., 2016 (30)	Rare diseases	154 families	Parents were satisfied with the services provided for diagnosis and follow-up, relationships with the family paediatrician, and rehabilitation services. Negative scores were reported for institutional and/or private support and family relationships in severe conditions.
Simpson *et al*., 2018 (31)	Adrenal insufficiency and congenital adrenal hyperplasia	14 mothers; 6 fathers	Parents in the UK reported the diagnosis period was characterised by a lack of awareness amongst healthcare professionals. Parents reported burden, anxiety and disruption associated with the intensive treatment regimen. Parents adjusted and gained confidence over time yet found delegating responsibility for medication difficult and worried about the future of their child. Access to psychological support and contact with other families was reported as highly beneficial.
Somanadhan *et al*., 2016 (32)	MPS Type I, II, III & VI	8 parents	Parents spoke of their child's QOL and the impact on their own physical and psychological wellbeing. Parents also reflected on issues of stigmatisation and isolation in their experience of living with a child with a rare disorder. Results indicated how caring for someone with MPS could impact all dimensions of the family's life. Parents gradually develop ways to incorporate MPS into their day-to-day life. The carers in this study experienced a range of uncertainties, with parents using terms such as 'no man's land' and 'future is unknown' to describe their world.
Toledano-Toledano & Luna 2020 (33)	Chronic illnesses	401 parents	Two profiles of caregivers of paediatric patients with chronic diseases were identified. Profile 1, called Vulnerability of family caregivers, is characterised by high levels of anxiety, depression, parental stress, and caregiver burden, accompanied by low levels of family support, resilience, and wellbeing. Profile 2, called Adversity of family caregivers, shows an inverse pattern, with high levels of family support, resilience, and wellbeing and low levels of anxiety, depression, parental stress, and caregiver burden.
Van De Loo *et al*., 2020 (34)	Mitochondrial disease	122 children and parents	Report revealed a lower QOL, more physical problems and behavioural issues compared to chronically ill patients. Tiredness and pain were frequently reported, although

			enhanced levels of stress regarding parenting were not reported, and parents experienced sufficient social support. At the end of the diagnostic process, 5.7% of the children received the genetically confirmed diagnosis of mitochondrial disease, 26% showed non-conclusive abnormalities in the muscle biopsy, 54% did not receive any diagnosis, and the remaining received other diagnoses. Strikingly, children without a diagnosis showed similar QOL and behavioural problems as children with a diagnosis.
Van Der Kloot *et al*., 2010 (35)	Sternocostoclavicular hyperostosis	52 patients	Patients reported posttraumatic stress symptoms, unfavourable illness perceptions, low health status, and poor QOL. Psychological distress in the pre-diagnostic period was associated with unfavourable conditions in the current situation.
Witt *et al*., 2019 (36)	Oesophageal atresia	63 parent-child dyads	Study showed that parent reports are a reliable source of information. Clinicians should therefore not only observe the paediatric patient but also the parents.
Wu *et al*., 2020 (37)	Epidermolysis bullosa	10 caregivers	Family caregivers experience many challenges when caring for epidermolysis bullosa patients and experience substantial stress and overwhelming burdens.
Yamaguchi *et al*., 2017 (38)	Inherited metabolic diseases	Review of 24 studies	Parents with infants suffered anxiety and distress because of the child's diet therapy and found that a parents' support group was essential. The psychological health of their caregivers was also poor.
Zengin *et al*., 2020 (39)	MPS	4 mothers; 4 fathers	This study determined that parents of children with an MPS diagnosis had many difficulties related to multi-systemic problems (orthopaedic problems, vision and hearing problems, speech disorders, cardiac problems) caused by the disease.
Zurynski *et al*., 2017 (40)	Rare diseases	462 children	Before receiving the correct diagnosis, 38% consulted ≥ 6 different doctors. Among those with a diagnosis, 37% believed the diagnosis was delayed, and 27% initially received a wrong diagnosis. Consequences of delayed diagnosis included anxiety, loss of reproductive confidence because of an ill-defined genetic risk, frustration, and stress (54%), disease progression (37%), delays in treatment (25%) and inappropriate treatments (10%). Perceived reasons for diagnostic delays included lack of knowledge about the disease among health professionals (69.2%), lack of symptom awareness by the family (21.2%) and difficulties accessing tests (17.9%). Some families (16%) were dissatisfied with the way the diagnosis was delivered, citing lack of empathy and lack of information from health professionals. Psychological support at diagnosis was provided to 47.5%, but 86.2% believed that it should always be provided.

LSD=lysosomal storage disease; MPS=mucopolysaccharidoses; pCNV=pathogenic copy number variations; QOL=quality of life.