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Social provisions in patients with mitochondrial diseases

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ABSTRACT

Background Mitochondrial diseases often follow a chronic, multimorbid disease course in adults. Like other chronic conditions, mitochondrial diseases present a challenge to public and community health models and patients are potentially at higher risk of social isolation and loneliness. However, there is lack of data on social provisions in mitochondrial diseases.

Methods We performed a cross-sectional observational study on patients with a confirmed genetic or clinical diagnosis of mitochondrial disease, recruited between September 2018 and December 2021. Participants completed the Social Provisions Scale (SPS) as a measure of social support. Designated carers similarly completed the SPS in carer-specific questionnaires.

Results 95 mitochondrial disease patients and 24 designated carers completed the SPS. Social provisions were met for all six subscales of SPS in the mitochondrial disease cohort: (1) guidance 90.5% (n=86), (2) reassurance of self-worth 82.8% (n=77), (3) social integration 88.4% (n=84), (4) attachment 83.2% (n=79), (5) opportunity of nurturance, 61.1% (n=58) and (6) reliable alliance 95.8% (n=91). All social provisions were also met in the carer cohort.

Conclusion Patients with mitochondrial diseases and their carers demonstrate a high perceived level of social support in the setting of a tertiary referral centre specialised in mitochondrial disease despite the burden of chronic disease.

INTRODUCTION

Mitochondrial diseases are multisystem disorders caused by pathogenic variants in mitochondrial DNA (mtDNA) or nuclear DNA (nDNA).[1] Patients often have a range of clinical manifestations and therefore, commonly experience multimorbidity, inclusive of more commonly recognised comorbidities. Adult patients with mitochondrial diseases may experience stepwise deteriorations and/or a chronic, progressive and complex clinical course.[2]

Chronic conditions (CCs) have been identified as 'an ongoing cause of substantial ill health, disability and premature death' contributing to 'global, national and individual health concern'.[3] People with CCs usually experience multimorbidity (ie, the presence of two or more CCs at the same time), often associated with complex health needs and poorer quality of life overall. Consequently, people with CCs are at a high risk of social isolation and loneliness[4 5] and are more severely impacted by a lack of social provisions than healthier individuals. There is a distinct lack of data on social support in mitochondrial diseases, which often present as CCs and are more prevalent than some other neurogenetic disorders.[6]

Social isolation and loneliness are serious public health concerns and are associated with negative health outcomes.[4 7] There are multiple definitions of social support, but the provision of support through interpersonal social relationships is paramount for an individual's perception of their quality of life.[8 9]

A number of social support measures have been developed including the Social Provisions Scale (SPS), which has been applied to people with disability and in CCs.[10] The SPS evaluates six domains of social provisions: (1) guidance (having people who can provide advice when needed), (2) reassurance of worth (having others validate one's value and competence), (3) social integration (sense of belonging to a group with common interests and social activities), (4) attachment (feelings of intimacy, peace and security), (5) opportunity for nurturance (providing care to others) and (6) reliable alliance (access to assistance in times of need from others).

While the SPS has been used for patients with neurological conditions like multiple sclerosis (MS)[11] and traumatic brain injury (TBI),[12] it has not been considered in mitochondrial diseases. To address this gap in understanding, we explored social provisions for patients with mitochondrial diseases.

METHODS

Participants for this cross-sectional observational study were recruited from the Mitochondrial Disease Clinic at Royal North

Shore Hospital in Sydney, Australia between September 2018 and December 2021.

Within part of a larger battery of questionnaires for a broader study, participants were asked to complete the SPS[10] considered here, as part of the questionnaires administered. The broader assortment of questionnaires was designed to answer research aims that are published elsewhere.[13 14]

The SPS is a 24-item scale that is divided into the six subscales described above. Each subscale comprises four items, of which two interrogate the presence of a social support and two interrogate the absence of a social support. The subscale scores for an absence of social support are inverted for calculating the sum of scores. Scoring is carried out at four levels of intensity: 1=strongly disagree, 2=disagree, 3=agree and 4=strongly agree. High scores indicate strong social relationships and low scores indicate a lack of social support that is, social isolation. Consistent with the prescribed SPS scoring method, we scored the sum total of items for each of the six subscales as follows:

1. Strongly agree >12 points
2. Agree=11 points
3. Indeterminate or 'Not Clear' = 10 points (mixture of agree and disagree)
4. Disagree=9 points
5. Strongly disagree <8 points

Social provisions were met if the score was greater than 10 points (ie, if the responses were strongly agree and/or agree).

Analyses

We report descriptive statistics, including frequency counts and percentages. We performed logistic regression analyses to determine any association of social provisions with the number of systems affected by mitochondrial diseases in the participants, living arrangements (alone or with someone) and the type of causative genetic variants (mtDNA or nDNA variants or nil variant detected). All statistical analyses were conducted using R and RStudio (R v.4.2.1).

RESULTS

99 participants were enrolled in the study with four participants not completing the SPS scale. The analysis presented

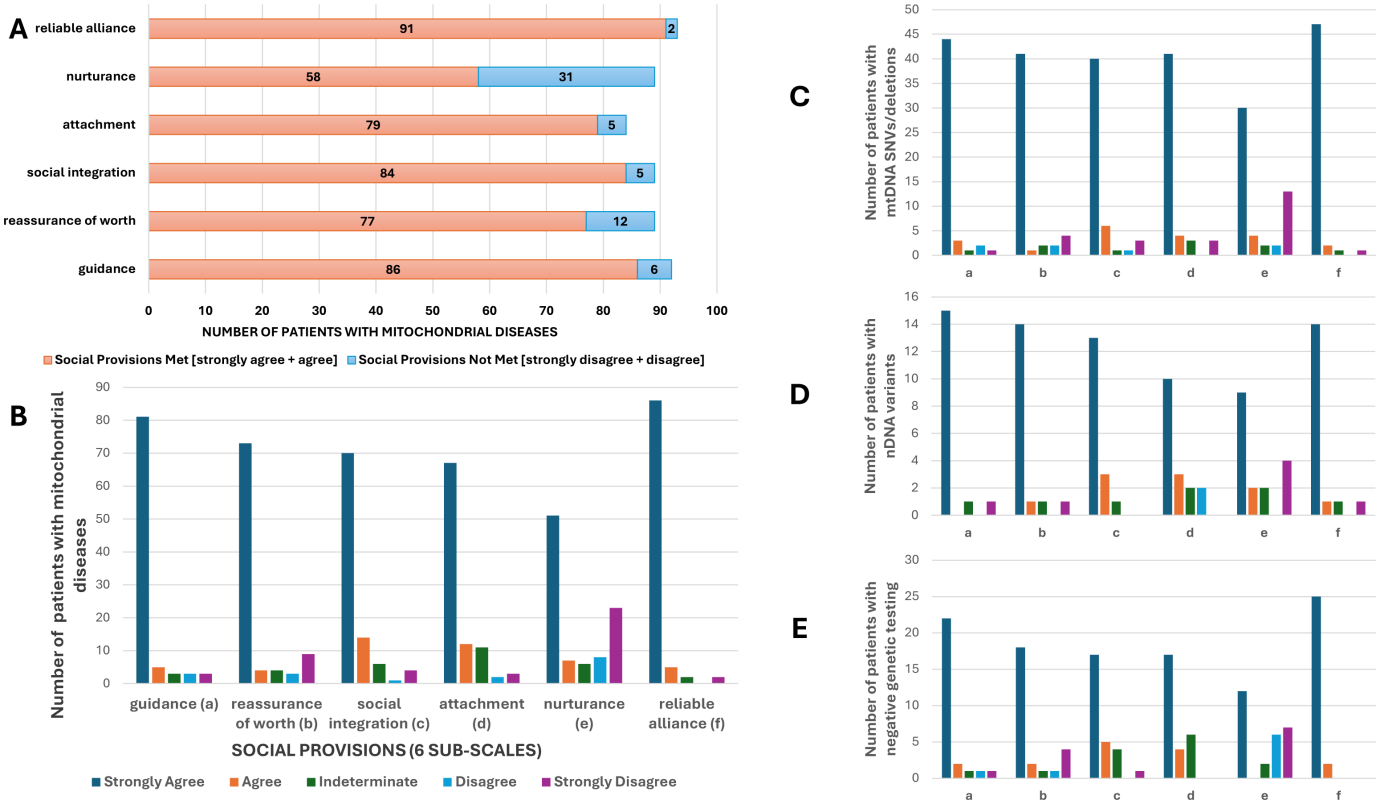

Figure 1 Distribution of social provisions in patients with mitochondrial diseases (n=95). **A**. The graph shows the number of patients in the study population who ▭, 'met' social provisions (strongly agree+agree) vs ▭, 'not met' social provisions (strongly disagree+disagree) for each of the six subscales. **B**. The graph shows the number of patients in the study population who scored ■, strongly agree; ■, agree; ■, indeterminate; ■, disagree and ■, strongly disagree for each of the six social provisions (guidance (a), reassurance of worth (b), social integration (c), attachment (d), nurturance (e) and reliable alliance (f)). The three vertically stacked graphs on the right display the distribution of these scores as subdivided by the genetic diagnosis of the patients: (**C**) social provisions in patients with mtDNA SNVs or deletions; (**D**) social provisions in patients with nDNA variants and (**E**) social provisions in patients with negative genetic testing. mtDNA, mitochondrial DNA; nDNA, nuclear DNA; SNVs, single nucleotide variants.

here considered the 95 participants who completed the SPS scale.

The mean age of participants at the time of recruitment was 53.7 years (SD 16.5). 66.3% of the cohort were females and 33.7% were males. Of the 95 participants, 51 had causative mtDNA single nucleotide variants (SNVs) or deletions and 17 had pathogenic variants in the nDNA. The remaining 27 participants did not have a molecular diagnosis after whole genome sequencing but fulfilled the clinical diagnostic criteria[15] and had muscle biopsy findings supportive of a clinical mitochondrial disease diagnosis. 74 participants lived with one or more family members. 24 participants had a designated carer (10 for participants with mtDNA SNVs or deletions, 8 for participants with nDNA variants and 6 for participants with inconclusive genetic testing). All carers of participants completed the SPS scale presented in carer-specific questionnaires.

Figure 1 shows the distribution of social provisions in the study population and is further subdivided by the genetic diagnosis of the participants. This cohort demonstrated strong social support, evident from a high frequency of scores consistent with strongly agree (≥12 points) across the six subscales of the SPS.

In our study population, 90.5% (n=86) indicated access to guidance, 82.8% (n=77) were reassured of self-worth, 88.4% (n=84) confirmed social integration, 83.2% (n=79) indicated attachment, 61.1% (n=58) had opportunity of nurturance and 95.8% (n=91) had a sense of reliable alliance. Meeting social provisions was similarly demonstrated in each of the diagnostic subgroups.

The sum of scores in each of the six subscales of the SPS completed by the 24 carers demonstrated that social provisions were also met for the carers. For access to guidance, it was 83.3% (n=20), for reassurance of self-worth, it was 95.8% (n=23), for social integration, it was 83.3% (n=20), for attachment, it was 87.5% (n=21), for opportunity of nurturance, it was 95.8% (n=23) and for the sense of reliable alliance, it was 87.5% (n=21).

Logistic regression modelling returned no evidence of an association between any of the six social provision subscales and the number of systems involved in patients with mitochondrial disease, living status (living alone or with someone) or the type of causative genetic variant.

DISCUSSION

This study is the first to examine social support and provisions in patients with mitochondrial diseases. We demonstrate that in the setting of our tertiary referral centre specialised in mitochondrial disease, all six social provisions in the SPS were clearly met across the cohort of participants and designated carers.

Studies have shown that CCs limit social participation and reduce the size of social networks. Additionally, stigma related to CCs undermines social interactions, resulting in a higher risk of social isolation that may precipitate loneliness.[4 5 16] Mitochondrial diseases in adults are considered CCs with a wide spectrum of severity.[1 2] Even patients with relatively mild symptoms at diagnosis may progress to a chronic multisystem disease pattern over their life span. With semiology consistent between mitochondrial diseases and CCs, it might be anticipated that a decline in one or more of the social provisions for the patients with mitochondrial diseases would be observed. Conversely, our study population displayed high perceived levels of social support for all six subscales of the SPS. Studies in MS and TBI have reported aggregate SPS scores with the conclusion that social provisions were met but at a level less than those individuals who had no disability.[11 12] These studies did not provide separate scores for the six social provisions, so we were unable to clarify the extent of these provisions for comparison with our cohort. Additionally, the generalisability of our findings to mitochondrial disease patients who are not treated within a tertiary referral centre is unclear.

As shown in figure 1, having reliable alliance was reported by the highest number of participants, followed by guidance, social integration, attachment, reassurance of worth and opportunity of nurturance. Compared with the other social provisions, opportunity of nurturance was satisfied by only 58 participants (61.1%), which may relate to the burden of comorbidities and physical disability limiting the patient's capacity to provide care to others, such as children or senior family members. It is understandable that the subscale of nurturance was clearly met for most carers (23 out of 24), as one of their major functions is to support a mitochondrial disease patient. The majority of carers were also satisfied for the other five social provisions.

The data did not demonstrate a statistically significant association between the six subscales of SPS and multimorbidity, living arrangements or the molecular diagnosis category. This could be attributed in part to the consistently high scores across all subscales for a substantial proportion of the cohort.

CONCLUSION

In the setting of our tertiary referral centre specialised in mitochondrial disease care, both patients and their carers demonstrated a high level of social support, consistent with well-supported interpersonal relationships among social ties. This indicates that provision and utilisation of opportunities for social interactions can be sustained throughout their disease course and justifies seeking care at a specialised tertiary referral centre regardless of the burden of chronic illness in these patients.

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Patient consent for publication Not applicable.

Ethics approval This study involves human participants. This study was approved by the Northern Sydney Local Health District Human Research Ethics Committee (NSLHD-HREC Reference number: LNR/17/HAWKE/268). Participants gave informed consent to participate in the study before taking part.

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REFERENCES

1 Davis RL, Kumar KR, Puttick C, *et al*. Use of Whole-Genome Sequencing for Mitochondrial Disease Diagnosis. *Neurology (ECronicon)* 2022;99:e730–42.
2 Gorman GS, Chinnery PF, DiMauro S, *et al*. Mitochondrial diseases. *Nat Rev Dis Primers* 2016;2:16080.
3 AIHW. *Chronic Conditions and Multimorbidity*. Australian Institute of Health and Welfare, 2022.
4 Christiansen J, Lund R, Qualter P, *et al*. Loneliness, Social Isolation, and Chronic Disease Outcomes. *Ann Behav Med* 2021;55:203–15.
5 Iovino P, Vellone E, Cedrone N, *et al*. A Middle-Range Theory of Social Isolation in Chronic Illness. *Int J Environ Res Public Health* 2023;20:4940.
6 Sue CM. Mitochondrial disease: recognising more than just the tip of the iceberg. *Med J Aust* 2010;193:195–6.
7 Leigh-Hunt N, Bagguley D, Bash K, *et al*. An overview of systematic reviews on the public health consequences of social isolation and loneliness. *Public Health (Fairfax)* 2017;152:157–71.
8 Chiu CY, Motl RW, Ditchman N. Validation of the Social Provisions Scale in people with multiple sclerosis. *Rehabil Psychol* 2016;61:297–307.
9 Wallston BS, Alagna SW, DeVellis BM, *et al*. Social support and physical health. *Health Psychol* 1983;2:367–91.
10 Cutrona CE, Russell D. The provisions of social relationships and adaptation to stress. In: Jones H, Greenwich (CT) PD, eds. *Advances in personal relationships*. . 1987: 1. 37–67.
11 Jaracz K, Pawlak M, Górna K, *et al*. Quality of life and social support in patients with multiple sclerosis. *Neurol Neurochir Pol* 2010;44:358–65.
12 McLean AM, Jarus T, Hubley AM, *et al*. Associations between social participation and subjective quality of life for adults with moderate to severe traumatic brain injury. *Disabil Rehabil* 2014;36:1409–18.
13 Haque S, Crawley K, Shrestha R, *et al*. Healthcare resource utilization of patients with mitochondrial disease in an outpatient hospital setting. *Orphanet J Rare Dis* 2023;18:129.
14 Haque S, Crawley K, Davis R, *et al*. Clinical drivers of hospitalisation in patients with mitochondrial diseases. *BMJ Neurol Open* 2024;6:e000717.
15 Morava E, van den Heuvel L, Hol F, *et al*. Mitochondrial disease criteria: diagnostic applications in children. *Neurology (ECronicon)* 2006;67:1823–6.
16 Halcomb E, Thompson C, Tillott S, *et al*. Exploring social connectedness in older Australians with chronic conditions: Results of a descriptive survey. *Collegian* 2022;29:860–6.