Psychosocial well-being and health-related quality of life in a UK population with Usher syndrome

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ABSTRACT

Objectives: To determine whether psychosocial well-being is associated with the health-related quality of life (HRQOL) of people with Usher syndrome.

Setting: The survey was advertised online and through deafblind-related charities, support groups and social groups throughout the UK.

Participants: 90 people with Usher syndrome took part in the survey. Inclusion criteria are having a diagnosis of Usher syndrome, being 18 or older and being a UK resident.

Primary and secondary outcome measures: All participants took part in a survey that measured depressive symptoms, loneliness and social support (predictors) and their physical and mental HRQOL (outcomes). Measured confounders included age-related, sex-related and health-related characteristics. Hierarchical multiple linear regression analyses examined the association of each psychosocial well-being predictor with the physical and mental HRQOL outcomes while controlling for confounders in a stepwise manner.

Results: After adjusting for all confounders, psychosocial well-being was shown to predict physical and mental HRQOL in our population with Usher syndrome. Increasing depressive symptoms were predictive of poorer physical (β=−0.36, p<0.01) and mental (β=−0.60, p<0.001) HRQOL. Higher levels of loneliness predicted poorer mental HRQOL (β=−0.20, p<0.05). Finally, increasing levels of social support predicted better mental HRQOL (β=0.19, p<0.05).

Conclusions: Depression, loneliness and social support all represent important issues that are linked with HRQOL in a UK population with Usher syndrome. Our results add to the growing body of evidence that psychosocial well-being is an important factor to consider in people with Usher syndrome alongside functional and physical impairment within research and clinical practice.

INTRODUCTION

The World Health Organisation defines health as a state of “complete physical, mental and social wellbeing.”1 A person’s health is a fundamental component of their well-being and satisfaction with their life.2 Quality of life (QOL) is a multidimensional and subjectively rated phenomenon that captures a person’s well-being and satisfaction with the myriad of biological, physical, social, psychological, spiritual and cultural influences on their life.3 4 There is a specific form of QOL related to health known as health-related quality of life (HRQOL). This measures the extent to which health is perceived to affect physical, psychological and social well-being.5

There is evidence that living with a combined visual and hearing impairment, also known as deafblindness, is associated with a reduced QOL.6 7 Researchers have posited what factors may link deafness and blindness with QOL. Factors such as functional impairments, social difficulties, role difficulties, communication issues, psychological factors and health have been posited to explain the reduced QOL in these populations.7–9 However, there is a lack of research that looks at what factors may predict a reduced HRQOL in people with deafblindness.

Psychological and social (psychosocial) well-being have been shown to predict QOL in the general population5 10 11 and in people with chronic illness.12–14 Existing evidence also tells us that deafblindness is linked to poorer psychosocial well-being. Deafblindness is linked to a high prevalence...
of social isolation and loneliness,15 increased psychological distress,16 depression,15 17 vulnerability,18 perceived stigma19 and perceived inadequate support.15 16 Therefore, psychosocial well-being may represent an important factor to consider in the HRQOL of people deafblindness.

One of the leading causes of deafblindness is the genetic condition Usher syndrome. Usher syndrome is a progressive chronic condition associated with retinitis pigmentosa (which causes visual impairment), sensorineural deafness (which causes hearing impairment) and, in some cases, vestibular dysfunction (which causes balance difficulties). There are three main subtypes of Usher syndrome: Usher 1, 2 and 3. These subtypes are differentiated by age of onset, severity of sensory impairments and presence of balance difficulties.

Health incorporates psychological and social well-being as well as physical well-being.1 Despite this, work into the health and well-being of people with Usher syndrome has typically been limited to examination of biological, physical and functional impairment.20 21 However, a growing body of research shows that it may also be important to also consider psychological and social well-being within this population.

Previous work has shown that Swedish adults with Usher syndrome type 1 and type 2 report more problems with depressive symptoms and higher suicidal ideation than the general population.24 25 Other studies have also shown that Usher syndrome in adults can be associated with stress, anxiety and depression.26–28 There is also evidence that Usher syndrome is linked to decreased social trust24 and increased feelings of social isolation and loneliness.28 However, there is also work showing the positive importance of social support to the lives of people with Usher syndrome.29

However, we do not yet know how psychosocial well-being may be linked to HRQOL in people with Usher syndrome. Much of the research on visual and hearing impairments with HRQOL examines either deafness or blindness alone.6 30–31 Or compares single sensory impairments with combined sensory impairments.7 Much of this work is also limited to older populations and does not examine Usher syndrome specifically.6 7 30

Several theories may explain why psychological well-being and HRQOL may be important to examine in people with Usher syndrome. We know that communication is an important aspect of everyday life and well-being. The challenges faced socially, in interacting with family and friends, having participation in the community and accessing and sharing the exchange of information in individuals with deafblindness may lead to a perceived reduction in HRQOL.31 Furthermore, research suggests that the constant adjustment experienced due to the deterioration and progressive nature of an illness such as Usher syndrome could lead to mental and emotional difficulties,32 such as depression.28 33

Therefore, the aim of this study was to determine whether psychosocial well-being is associated with physical and mental HRQOL in a UK-resident population of adults with Usher syndrome.

**METHODS**

**Participants**

A total of 90 eligible participants completed the survey between September 2015 and February 2016 (for participant characteristics, see table 1). Inclusion criteria were a self-reported diagnosis of Usher syndrome, aged 18 or older and resident in the UK. Participants were recruited through convenience and snowball sampling. The study was promoted using social media (with linked in British Sign Language YouTube video), meetup groups, emails and magazine advertisements through the following charities and groups: Sense UK, Sense Northern Ireland, Sense Usher service team, Deafblind UK, Deafblind Scotland, UsherVibe and The Limping chicken website.

In total, 120 people showed an interest in completing the study; however, 15 did not meet inclusion criteria. The completion rate was 86% with 10 people who made
initial contact opting not to take part and 5 not completing the survey.

**Study design**
The predictor variables were depressive symptoms, loneliness and social support. The outcome measures were physical and mental HRQOL. Measured confounders included age-related, sex-related and health-related characteristics.

**MATERIALS**

**Outcome measure: HRQOL**
The 12-Item Short-Form Health Survey V.2 (SF-12v2)
This 12-item survey measures HRQOL over 4 weeks and is based on the longer SF-36 Health Survey. The scale is a validated and reliable tool that has been used globally. The scale contains questions that examine eight domains of HRQOL: physical functioning, role (physical), bodily pain, general health perceptions, vitality, social functioning, role (emotional) and mental health.

The SF-12v2 can also be used to calculate composite scores for physical HRQOL (physical component score: PCS) and mental HRQOL (mental component score: MCS). Scores were calculated using the validated standardised norm based scoring algorithms for the PCS and MCS. All scores ranged from 0 to 100, with 50 representing the standardised norm score. This means that a score <50 indicates lower than the standardised average for HRQOL.

**Predictor variables**

**Participant characteristics**

**Sociodemographic characteristics**
Data were collected about gender (male/female), age (18–25, 26–35, 36–45, 46–55, 56–65, 66+) and occupational status (employed/self-employed, unemployed, student, retired) were collected.

**Health-related characteristics**
Self-rated questions on health were asked, including asking participants their Usher type (Usher 1, 2, 3, unknown). Level of hearing loss was assessed by asking participants which of the following (mild, moderate, severe/profound, unknown) best described their level of deafness. Categories used are those used within UK severe/profound, unknown) best described their level of deafness. Participants were also asked to identify their sight registration status (partially sighted, blind/severely sight impaired, unknown). This was based on UK categories for the registration of impaired sight. Finally participants were asked a single self-rated question about whether they had any other disabilities and health illnesses (yes or no).

**Psychosocial well-being characteristics**

Patient Health Questionnaire Mood Scale (PHQ-9)
Depressive symptoms were assessed with the PHQ-9 a widely used, validated and reliable screening tool for depression. The signed version of this questionnaire has also been validated for use in deafblindness. This questionnaire screens symptomatology of the nine depression symptoms used in the Diagnostic and Statistical Manual over the last 2 weeks.

Each item was scored from not at all (0), to nearly every day, with possible total scores ranging from 0 to 27 (the higher the score the higher the depressive symptoms). While the continuous score was used within our analyses, a score of 10 or more is indicative of clinically significant depressive symptomatology.

**The 3-item UCLA-loneliness scale**
The 3-item UCLA-loneliness scale measures loneliness with three items taken from the widely used 20-item revised UCLA-loneliness scale. The three items are: How often do you feel you lack companionship? How often do you feel isolated from others? How often do you feel left out?

Each question can be answered hardly ever/never (scoring 1), some of the time (scoring 2) and often (scoring 3) with scores ranging from 3 to 9 and higher scores indicative of higher loneliness. The use of the 3-item UCLA-loneliness scale has been validated in the English Longitudinal Study of Ageing studies.

**The 8-item Modified Medical Outcomes Study Social Support Survey (mMOS-SS)**
The mMOS-SS was used to collect data on levels of social support. This is an 8-item short-form version of the original widely used 19-item Medical Outcomes Study Social Support (MOS-SS). The scale is valid and reliable in measuring social support in health conditions.

Responses for each question range from a score of 1 to 5. The total score for all the questions are then calculated as the mean score and transformed to a standardised 0–100 scale. The higher the overall total score, the more social support.

**Questionnaire format**
The survey was made available in paper, electronically, online, telephone and Skype or face-to-face structure questionnaire interview formats to best meet the wide ranging and variable visual and hearing needs of each individual with Usher syndrome and provide equal access to the study. The provision of a qualified and experienced interpreter in British Sign Language (BSL) was used in four Skype and deafblind hand on signing (where signing was conducted through touch on the participants body) was used in one face-to-face structured questionnaire interview. The same interpreter was used for all interviews.
Ethical approval
Prior to taking part in the study, all participants gave their informed consent by either signing a consent form, giving verbal or signed consent if this was not possible, or checking an online item after reading the information sheet or having the information sheet signed to them where appropriate.

Data set
The data set can be found through Brunel University London figshare.47

Analysis
Descriptive analyses (frequencies, percentages and SDs) were used to describe all predictors, outcomes and confounders. Pearson’s correlational analyses were undertaken to explore the relationship of predictors and outcomes. Finally, two hierarchical multiple linear regression models were run to investigate the association of each psychosocial well-being predictor (depressive symptoms, loneliness and social support) with the PCS and MCS. All analyses were initially run unadjusted (model 1) and then were adjusted in a stepwise manner for sociodemographic variables (age and sex (model 2)), model 2+health-related characteristics (Usher type, other disability/illness, level of hearing loss and sight registration status (model 3)), model 3+other psychosocial well-being predictors (model 4). A power calculation indicated that to have a moderate effect size with a power of 0.8, at an α level of 0.05 with nine predictors that a sample size of 80 would be sufficient. All data were checked for normality (as indicated by a non-significant result in the Shapiro-Wilk test) and multicollinearity (as assessed with a correlation matric and variance inflation factors) prior to running the analyses and all assumptions were met for running the multiple regression. Analyses were carried out with SPSS V.20.0.

RESULTS
Descriptive statistics and correlational analyses
Of the 90 participants completing the study, the majority of participants were aged 36–45 and females (see table 1). Furthermore, most participants presented with Usher type 2, reported having severe hearing loss and being registered blind or having a severe sight impairment (see table 1).

The mean values of psychosocial well-being and HRQOL variables indicated the physical and mental HRQOL were lower than the standardised mean (see table 2). Results also indicated the mean depression score was close to the cut-off of 10 normally taken to indicate high depressive symptoms (see table 2). The mean loneliness score indicated moderate levels of loneliness and the mean social support score indicated a higher than average level of social support (see table 2).

Correlational analyses indicated a negative relationship between physical HRQOL with depressive symptoms (see table 3). They also indicated a negative relationship between mental HRQOL with depressive symptoms and loneliness. However, mental HRQOL was positively related to social support (see table 3). In addition, there was a positive correlation between depressive symptoms with loneliness, and a negative correlation between loneliness with social support (see table 3).

Psychosocial predictors of physical HRQOL
The hierarchical linear regression model indicated that depressive symptoms were significantly associated with physical HRQOL (see table 4). The association remained significant after controlling for all confounders. However, neither loneliness nor social support was significantly associated with physical HRQOL (see table 4). The fully adjusted model that included all sociodemographic, health-related and psychosocial predictors explained 43% of the variance in physical HRQOL (R²=0.45). Those variables that were most significantly

Table 2  Psychosocial well-being and HRQOL-related descriptive statistics

<table>
<thead>
<tr>
<th>Measure</th>
<th>Questionnaire</th>
<th>Number (frequency, %)</th>
<th>Mean (SD)</th>
</tr>
</thead>
<tbody>
<tr>
<td>HRQOL</td>
<td>SF-12 PCS</td>
<td>≥50 (&gt; standardised mean)</td>
<td>27 (30.0)</td>
</tr>
<tr>
<td></td>
<td>&lt;50 (&lt; standardised mean)</td>
<td>58 (64.4)</td>
<td></td>
</tr>
<tr>
<td>Depressive symptoms</td>
<td>PHQ-9</td>
<td>≥50 (&gt; standardised mean)</td>
<td>25 (27.8)</td>
</tr>
<tr>
<td></td>
<td>&lt;50 (&lt; standardised mean)</td>
<td>65 (72.2)</td>
<td></td>
</tr>
<tr>
<td></td>
<td>&lt;10 (low depressive symptoms)</td>
<td>56 (62.2)</td>
<td>9.38 (7.1)</td>
</tr>
<tr>
<td></td>
<td>≥10 (high depressive symptoms)</td>
<td>34 (37.8)</td>
<td></td>
</tr>
<tr>
<td>Loneliness</td>
<td>3-item UCLA-loneliness scale</td>
<td>6.09 (1.8)</td>
<td></td>
</tr>
<tr>
<td>Social support</td>
<td>MOSS-SS</td>
<td>58.9 (22.9)</td>
<td></td>
</tr>
</tbody>
</table>

The table shows mean values for the HRQOL outcome measures and psychosocial predictor variables. For the PCS and MCS, a score <50 indicates that HRQOL for each area is lower than the standardised mean (ie, worse than normal). For the PHQ-9, a score of 10 or more is indicative of clinically significant depressive symptomatology. There are no defined cut-offs for the 3-item loneliness scale or the MOSS-SS and so frequencies were not included for these questionnaires.

HRQOL, health-related quality of life; SF-12, short-form 12-item; MSC, mental component score; PCS, physical component score; PHQ-9, patient health questionnaire 9-item; MOSS-SS, medical outcomes social survey-social support.
This study provides evidence that psychosocial well-being is linked to HRQOL in a UK population with Usher syndrome. Depressive symptoms were predictive of poorer physical and mental HRQOL. Loneliness was predictive of poorer mental HRQOL and social support predictive of better HRQOL. These results provide preliminary evidence that psychosocial well-being may be an important consideration for the HRQOL of people who have Usher syndrome.

### Psychosocial predictors of mental HRQOL

The hierarchical multiple linear regression model showed that depressive symptoms, loneliness and social support were all independently associated with mental HRQOL (see table 5). All relationships remained significant after controlling for confounders. The fully adjusted model that included all sociodemographic, health-related and psychosocial predictors explained 61% of the variance in mental HRQOL ($R^2=0.61$). Those variables that were most significantly associated with mental HRQOL were depressive symptoms, loneliness and social support (see online supplementary appendix I). Depressive symptoms ($\beta=-0.60$, $p<0.001$) and loneliness ($\beta=-0.20$, $p=0.05$) were associated with poorer HRQOL. Whereas social support was associated with better mental HRQOL ($\beta=0.19$, $p=0.05$) (see online supplementary appendix I for fully adjusted model).

### Depressive symptoms and HRQOL

Previous work has shown that people with Usher syndrome have a high self-reported level of depression.25 Furthermore, previous work indicates that there are high levels of suicidal ideation in people with Usher syndrome types 1 and 2.24 25 28 33 Our work adds to this growing body of research by indicating that not only is the prevalence of depression high in this population, but it also has a negative association with physical and mental HRQOL.

Previous work has hypothesised that depression in people with deafblindness is linked to a myriad of problems with communication, reduced physical and functional activity, difficulties interacting with others and low satisfaction with their social activities.28 32 33 48 These kinds of communication, functional and social issues have been hypothesised to explain the link between deafness and blindness with reduced QOL.7-8 Thus, it is possible that the link between depressive symptoms with reduced HRQOL could be due to the link between depressive symptoms with reduced physical, function,
communication and social well-being. Future work should determine how physical, functional and communication difficulties are linked with depressive symptoms and how these are associated with HRQOL.

**Loneliness and HRQOL**

Previous research has suggested that the poor psychological well-being of people with dual sensory impairment is due mostly to their experience of social isolation. Our results indicate that as levels of loneliness increase that the mental HRQOL of our population decreased.

Social well-being and feeling connected with people are fundamental components of QOL. Qualitative studies conducted in people with Usher syndrome have shown that feelings of loneliness are linked to feeling isolated and lack of social support. Other research also indicates that loneliness is linked with higher depressive symptoms in people with deafblindness. Results from our study also provide additional evidence that loneliness is correlated with higher depressive symptoms and lower social support in people with Usher syndrome. Future work should examine the relationship of loneliness with other indicators of psychosocial well-being in Usher syndrome.

Feelings of loneliness could also result in part from difficulties in communication. Previous work in hearing impaired populations and people with Usher syndrome type 1 indicates that ability to communicate and improved hearing are linked with improved HRQOL. Thus, it is possible that loneliness and isolation could result in part from difficulties in communication. Future work should determine how problems with communication in Usher syndrome are linked to loneliness and HRQOL.

**Social support and HRQOL**

Support from family, friends and healthcare professionals is a fundamental component of QOL. There is also a large amount of work demonstrating the importance of social support for health. Previous work has also shown that social support is associated with less disability-related distress and limitations in activities of daily living and improved psychosocial well-being. Conversely, having less social support can be linked to poorer mental well-being. Thus it is perhaps not surprising that our results indicate that increasing social support is predictive of improved mental HRQOL in people with Usher syndrome.

Qualitative research has also emphasised the positive importance of social support for people with Usher syndrome. In one study, participants emphasised that emotional support and companionship was more important than help with practical issues. Our work adds to this by demonstrating that social support is also linked with a better mental HRQOL. Future work should determine how social support is linked with improved HRQOL in people with Usher syndrome, and whether social support interventions such as peer-led support could be used to help improve HRQOL in this population.

**Strengths and limitations**

To the best of our knowledge, this is the first study that has examined the association of psychosocial well-being with HRQOL in a population with Usher syndrome. Another notable strength of this study is the wide range of ages of people who took part. Most psychosocial research on deafblindness is in older populations, thus results from this study can be extrapolated outside of older populations. However, our sample was largely employed, female and has Usher type 2, which is not necessarily representative of the UK Usher syndrome population. The sampling method employed was opportunistic, and primarily performed through the internet and support groups. This could mean that our sample is not necessarily representative of a community-based Usher syndrome population (ie, it is possible that more highly educated and functional participants who are more engaged with support groups would be internet users and participate in the community-organisations...
that we primarily recruited through). These issues of potential bias and generalisability should be borne in mind when interpreting results.

Another limitation that should be acknowledged is that many of the measures used were shorter versions of more lengthy questionnaires. For example, the 3-item UCLA-loneliness scale is based off the longer 20-item scale, and while validated this scale may not capture the full complexity and intricacies of the loneliness experienced within our sample. Furthermore, the measure of depression was not a clinical measure of depression. There may be additional issues with the self-report items used to measure the level of hearing loss and sight registration status and the non-specific question on additional illness/disability. These questions do not fully capture illness severity of comorbidities or tell us much about what kinds of other illnesses/disabilities people were experiencing. However, we opted to include shorter measures of questionnaires (eg, SF-12 instead of the more detailed SF-36) and short questions in order to create a questionnaire that would not be too lengthy for this population to complete. Owing to the difficulties this population can have with reading material, we wanted to make sure that we could maximise the information we could collect while being sensitive to creating a questionnaire that would not be too long or difficult to complete. A possible final issue with the survey was that while validated questionnaires were used, that some have not been validated in a population with Usher syndrome. However, the PHQ-9 was validated for use in deafblind populations and the UCLA-loneliness scale has been used previously in deaf and blind populations.

There was also the issue that many people did not know what Usher syndrome they had or their sight registration status. As categorisation of visual impairment status was based on categories for sight registration status, this meant many participants may not have had their sight difficulties registered. There were also a large number of people who did not know their Usher type, while it is not clear why this is; it could be interesting to explore this issue in future research. This limitation means that we may not have had the statistical power to detect between-group differences for these variables with HRQOL.

In total, 90 people took part in the study, which is a relatively small sample size. However, the prevalence of Usher syndrome in the UK is relatively low with an estimated 9750 people diagnosed, meaning that a sample of 90 could be seen to be a good sample size for this population.

Finally, this was a cross-sectional study that limits inferences on causality. Future longitudinal work will be necessary to elucidate directionality of association between psychosocial well-being and HRQOL.

Clinical implications

Our results in tandem with other psychosocial research which shows the negative impact of Usher syndrome on well-being and HRQOL indicate the importance of health and social care professionals considering the importance of psychosocial well-being in this population. It is recommended that those professionals who work with this population should consider routinely screening for psychosocial well-being alongside monitoring physical health. It is also suggested that discussions around social support should also take place as higher levels of social support are linked with improved HRQOL.

It will also be important that healthcare professionals be mindful of the importance of themselves as sources of social support for people with Usher syndrome and other sensory impairments. In order to ensure that they are supporting people to the best of their abilities they should make sure all support and screening for psychosocial well-being is accessible to people with hearing and visual impairments in line with Department of Health recommendations.

Future directions

Future work could take a broader approach to examining HRQOL in this population in order to determine what physical, psychological and social factors are most predictive of HRQOL in people with Usher syndrome. Important additional factors to consider in future work could include physical and functional issues such as illness severity, functional limitations and communication issues. By including these we could get a better overall picture of what factors best predict physical and mental HRQOL in this population.

It will be important to conduct longitudinal studies that examine the direction of causality between psychosocial well-being and HRQOL. By conducting such studies we can know where to target possible interventions that will improve the HRQOL of people with Usher syndrome in the future. To the best of our knowledge, there is no study that has examined psychosocial well-being and Usher syndrome longitudinally. Owing to the complex and deteriorating nature of this condition, it will be important to determine how illness progression affects people’s psychosocial well-being.

CONCLUSIONS

Results from this study provide the first evidence that psychosocial well-being in Usher syndrome is associated with HRQOL in a UK population with Usher syndrome. This work adds to a growing area of research that is showing the importance of considering psychosocial well-being in people who have Usher syndrome. Future work can determine more broadly how physical, social, functional and social factors interact to affect the HRQOL of people with Usher syndrome so that interventions to help improve the HRQOL of this population can be improved.

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Contributors GD collected the data with help from AM and RS. AO and KJS prepared the data file. KJS and GD analysed the data and wrote the manuscript. All authors reviewed and edited the final manuscript for publication.

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