Disability and Rehabilitation

ISSN: 0963-8288 (Print) 1464-5165 (Online) Journal homepage: https://www.tandfonline.com/loi/idre20

Rare diseases: matching wheelchair users with rare metabolic, neuromuscular or neurological disorders to electric powered indoor/outdoor wheelchairs (EPIOCs)

Lorraine H. De Souza & Andrew O. Frank

To cite this article: Lorraine H. De Souza & Andrew O. Frank (2016) Rare diseases: matching wheelchair users with rare metabolic, neuromuscular or neurological disorders to electric powered indoor/outdoor wheelchairs (EPIOCs), Disability and Rehabilitation, 38:16, 1547-1556, DOI: 10.3109/09638288.2015.1106599

To link to this article: https://doi.org/10.3109/09638288.2015.1106599

Published online: 30 Dec 2015.

DISABILITY AND REHABILITATION, 2016
VOL. 38, NO. 16, 1547–1556
http://dx.doi.org/10.3109/09638288.2015.1106599

Taylor & Francis
Taylor & Francis Group

RESEARCH PAPER

Rare diseases: matching wheelchair users with rare metabolic, neuromuscular or neurological disorders to electric powered indoor/outdoor wheelchairs (EPIOCs)

Lorraine H. De Souza[a]* and Andrew O. Frank[a,b]

[a]Centre for Research in Rehabilitation, College of Health and Life Sciences, Mary Seacole Building, Brunel University London, Uxbridge, Middlesex, UK; [b]Stanmore Specialist Wheelchair Service, Royal National Orthopaedic Hospital, Brockley Hill, Stanmore, UK (Frank)

ABSTRACT

Purpose: To describe the clinical features of electric powered indoor/outdoor wheelchair (EPIOC) users with rare diseases (RD) impacting on EPIOC provision and seating. *Method*: Retrospective review by a consultant in rehabilitation medicine of electronic and case note records of EPIOC recipients with RDs attending a specialist wheelchair service between June 2007 and September 2008. Data were systematically extracted, entered into a database and analysed under three themes; demographic, diagnostic/clinical (including comorbidity and associated clinical features (ACFs) of the illness/disability) and wheelchair factors. *Results*: Fifty-four (27 male) EPIOC users, mean age 37.3 (SD 18.6, range 11–70) with RDs were identified and reviewed a mean of 64 (range 0–131) months after receiving their wheelchair. Diagnoses included 27 types of RDs including Friedreich's ataxia, motor neurone disease, osteogenesis imperfecta, arthrogryposis, cerebellar syndromes and others. Nineteen users had between them 36 comorbidities and 30 users had 44 ACFs likely to influence the prescription. Tilt-in-space was provided to 34 (63%) users and specialised seating to 17 (31%). Four users had between them complex control or interfacing issues. *Conclusions*: The complex and diverse clinical problems of those with RDs present unique challenges to the multiprofessional wheelchair team to maintain successful independent mobility and community living.

➤ IMPLICATIONS FOR REHABILITATION

- Powered mobility is a major therapeutic tool for those with rare diseases enhancing independence, participation, reducing pain and other clinical features.
- The challenge for rehabilitation professionals is reconciling the physical disabilities with the individual's need for function and participation whilst allowing for disease progression and/or growth.
- Powered wheelchair users with rare diseases with a (kypho) scoliosis require a wheelchair system that balances spine stability and movement to maximise residual upper limb and trunk function.
- The role of specialised seating needs careful consideration in supporting joint derangements and preventing complications such as pressure sores.

ARTICLE HISTORY
Received 3 August 2015
Revised 22 September 2015
Accepted 7 October 2015
Published online
18 November 2015

KEYWORDS
Assistive technology, clinical features, comorbidity, Friedreich's ataxia, powered mobility, rehabilitation

Introduction

Rare diseases (RD) are conditions affecting less than five in 10 000 of the general population,[1] or a prevalence of fewer than 200 000 affected individuals in the United States.[2] There are nearly 7000 rare diseases [3] with birth prevalence ranging from 450.0 per 100 000 to one recorded case worldwide.[4] Often RDs have no treatment, or ineffective treatment and are known to be very complex.[3] Many are life-threatening or chronically debilitating diseases, often of genetic origin.[1]

Genetics has greatly enhanced our understanding of the cause and nature of many RDs. However, the strategic documents relating to the management of

*Stanmore Specialist Wheelchair Service has now been disbanded.

CONTACT Lorraine H. De Souza lorraine.desouza@brunel.ac.uk Centre for Research in Rehabilitation, College of Health and Life Sciences, Mary Seacole Building, Brunel University London, Uxbridge, Middlesex, UK

RDs lack any reference to rehabilitation strategies that could ameliorate some of the disabling consequences of RDs,[5–11] and these reports make no reference to the functional impact on individuals with these disorders experiencing progressive disabilities. Furthermore, there is no reference to the inclusion of wheelchair provision as a means to promote participation and improve quality of life (QOL).

Those with RDs progressing to severe mobility impairments do not comprise a homogeneous group. Generally, they fall into three groups comprising metabolic dysfunction, for example, Morquio's disease, neuromuscular/neurological conditions, for example, Dejerine–Sottas disease and connective tissue/bone disorders, for example, osteogenesis imperfecta. Some features of these conditions present rehabilitation professionals with unusual challenges, including multiple fractures, multiple contractures or skeletal malformations, dwarfism and skin affectations in addition to the more commonly recognised issues of scoliosis and problematic pain. The majority of those with metabolic dysfunction and connective tissue/bone disorders do not experience cognitive deterioration as part of the disease progression but those with neuromuscular/neurological conditions may develop cognitive problems, for example, motor neurone disease (MND) [12] and progressive supranuclear palsy (PSP) [13] that may influence ability to drive EPIOCs.

Some RDs with larger numbers do have rehabilitation pathways and recognised mobility disability requiring wheelchair provision, for example MND [14,15] and Friedreich ataxia.[16] For less commonly seen RDs, case reports can provide some knowledge about wheelchair prescription.[17] Patient experience reports [18] and patient surveys [19] may be helpful, while disease registers may give insights into wheelchair use.[20] The therapeutic uses of powered wheelchairs, as reported for those with multiple sclerosis [21] have not been reported in RDs to our knowledge.

In the United Kingdom, the National Health Service (NHS) provides funded electric powered indoor/outdoor wheelchairs (EPIOCs) to people with severe and complex disabilities who fulfilled strict criteria.[22] Those eligible are unable to walk around their home unaided, self-propel and are able to utilise the chair independently. Scooters are not provided by the NHS. Users may choose to take the value of the prescribed EPIOC in vouchers and purchase a chair privately.[22,23]

The consideration of wheeled and/or powered mobility can provide substantial improvements to QOL.[24] The literature on wheelchair use in RDs appears negative in nature – in that a condition had deteriorated such that a wheelchair was needed for mobility.[25–29] The UK rare disease strategy makes no reference to mobility disability and the need for a full disability assessment and rehabilitation.[7] This is surprising in view of the proven benefits of powered mobility to the well-being of electric powered indoor/outdoor powered wheelchairs (EPIOC) users (referred to as 'users'), particularly the psychological and functional gains experienced by younger users [30] and the importance attributed by service users to participation.[31]

Previous research has identified the wide range of diagnoses, age and associated clinical features of recipients of EPIOCs. This research commented on the complex interactions between the chair user and the technical features of EPIOC prescription.[32] Users with RDs presented unique challenges to service providers and there is a paucity of evidence to inform clinical decisions. It has been recommended that more research is needed into management of patients with RD to underpin the development of guidelines to improve care.[7] Consequently, this study explores a subgroup, diagnosed with RDs, of a larger cohort with severe mobility disability.[32]

Similar to other groups of very severely disabled individuals, those with RDs face issues of ageing with a disability as well as the continuing trajectory of their condition, which for many will be deteriorating.[33] It is also recognised that they will experience secondary pathologies with accelerated age-related conditions or comorbidities.[34] Powered mobility is recognised to improve access and autonomy of persons ageing with a disability and it has been emphasised that understanding the specific needs of individuals is required to adapt assistive technologies in a way that is beneficial and useable.[34] 'Recognising the patients' individual symptomatic pattern of comorbidity' [35] is seen to be critical for analyses that extend beyond the diagnostic label to 'improve health status'.[35]

Therefore, the aim of this cross-sectional study is to describe those demographic and clinical features of people with RDs that impact on EPIOC provision and seating needs and to explore the complexities of comorbidities, features of RDs and conditions secondary to disability that impact on powered wheelchair provision and clinical management. Because many RDs are present from birth, a further aim was to determine whether age influenced the prescription of seating and chair features.

Methods

This is a cross-sectional study of a clinic population with retrospective review of electronic and case note records.

The setting

The Specialist Wheelchair Service at Stanmore was set up in 1997 [22] to provide a regional service for around 3.1 million people from both rural and inner city areas. Provision was limited to those who were unable to walk safely around their home, unable to self-propel and were judged safe to use their chairs in public places irrespective of age, diagnosis or time using a wheelchair (if any). The full eligibility criteria have been published.[22]

Provision involved:

- Completion of a screening questionnaire.
- Occupational therapy assessment for the suitability of the home environment and the likelihood that the eligibility criteria would be fulfilled.
- Children were assessed by their paediatric therapist to provide details of current management and an evaluation of cognitive, emotional, visuospatial and physical development relating to their suitability for EPIOC driving.
- Assessment at the multiprofessional (as recommended [36]) specialist regional service including eye and physical examination to define any problems with seating or controlling a powered wheelchair, concluding with a driving assessment to ensure satisfactory control of the wheelchair and safety for the users and others.
- A rehabilitation engineer delivered the wheelchair and explained its use, checked seating and that driving appeared satisfactory.

Participants

Potential participants lived in the community and were referred from their local wheelchair service to the specialist regional service which decided provision of an EPIOC based on clinical grounds. Inclusion criteria for this study were all individuals, who had been prescribed an EPIOC, were currently using their chair and had a diagnosis of a RD defined as a condition affecting less than five in 10 000 of the general population,[1] of metabolic, neurological or neuromuscular origin and recorded as the main diagnosis for ten or fewer individuals. Exclusions were those who did not fulfil the eligibility criteria for NHS EPIOC use.[22]

Data collection

Data had been recorded in two main sources. Firstly, the electronic record contained personal, demographic and diagnostic information. EPIOC prescriptive features included use of special seating (SS) (adaptive seating), tilt-in-space (TIS) and complex controls. Demographic data, diagnosis and wheelchair factors had been entered into the electronic record by health professionals after a multiprofessional physical assessment and examination. Secondly, patient notes (charts) contained clinical details relevant to the EPIOC provided.

Both records were reviewed between June 2007 and September 2008 by a consultant physician in rehabilitation medicine who was responsible for all patient care. Data were systematically extracted and entered into a computer database for analysis and all data anonymised.

Demographic profiles consisted of information on age and gender at initial assessment. Clinical profiles included: primary diagnosis, comorbidities, other clinical features and complications relating to the disability. Wheelchair factors included information about SS, defined as 'that which is needed by people who require a wheelchair but due to instability or deformity need additional support in order to function'.[37] Other data included TIS, cushions and complex controls.

Methods of analysis

Data were analysed to describe proportions and frequencies of variables relating to wheelchair features and SS provision. Comorbidities (conditions with no known or unlikely association with the index diagnosis), features of the RD and features of disability were categorised by type of description and by frequency of occurrence. Descriptive statistics were used to analyse demographic data (age and sex). Clinical issues were categorised into major diagnosis contributing to the need for a wheelchair and whether it was inherited (autosomal dominant, recessive or X-linked).

Data were analysed using *t* tests for significant differences in age between those users with SS or TIS and without.

This study was approved by the National Research Ethics Service.

Results

Fifty-four EPIOC users, mean age 37.3 (SD: 18.6; range: 11–70) years met the inclusion criteria. There were 27 males mean age 36 (SD: 17.7; range: 11–68) years and 27 females mean age 38.7 (SD: 19.8; range: 13–70) years. The incidence or prevalence of their condition (where known) are given in Table 1 and their diagnoses and clinical features are given in Table 2.

The majority of users had neurological conditions ($n = 31$) of which 10 had Friedreich's ataxia (five men, five women mean age 29.1, range 16–43, SD 11.0 years) and six had motor neurone disease (five men, one

woman mean age 58, range 51–63 years). A further three had neuromuscular conditions (central core disease, dystrophia myotonica and congenital myasthenia. Twenty users had disorders involving connective tissue and 42 users had inherited conditions, including two sisters both with infantile systemic hyalinosis (Table 2).

Comorbidities and additional clinical features

Sixteen users (30%) had no comorbidities or ACFs (Table 2). Nineteen users had between them 35 comorbidities and 31 users had a total of 45 ACFs (Table 2). Back pain was a common comorbidity ($n = 7$) and one user had additional neck pain. Six users had three or more comorbidities.

Hypertension was reported in five users. Scoliosis was a frequent ACF ($n = 8$), as was problematic pain ($n = 10$), often associated with other ACFs (Table 2). Four users had three or more ACFs. Eight users had needed orthopaedic surgery prior to EPIOC provision.

Wheelchair features

TIS was provided to 34 (63%) users and SS to 17 (31%) (Table 2). Six users had individually tailored seating systems. Carved foam seating was provided to three (Morquio's with cervical and spinal fusions, Friedreich's ataxia with a scoliosis and infantile systemic hyalinosis with severe scoliosis and fragile skin), Caps II to a user with Krabbe's disease, Matrix seating to a user with osteogenesis imperfecta and one user with Pelizaeus–Merzbacher disease was provided with a moulded seat insert. All other users needing SS were provided with appropriate standard cushions. Only three users who were provided with SS did not have one or more ACFs. TIS was provided to all eight users with scoliosis and SS to six with scoliosis.

Those provided with SS were significantly younger than those who had standard equipment ($p < 0.004$). There was no significant difference in age between those provided with TIS and those without.

Complex controls

Four users had between them complex controls (3), interfacing issues (2) and were tray mounted (2). A male aged 26 with osteogenesis imperfecta and comorbid asthma was provided with a tray mounted non-standard control system that needed to interface with other equipment. He required matrix seating but not TIS. A 16-year-old female user with infantile systemic hyalinosis complicated by scoliosis and poor skin condition needed extra sensitive complex controls, SS and TIS. A 23-year-old male with familial spastic paraplegia needed a tray mounted complex control and SS. A 20-year-old female with Krabbe's disease needed controls interfacing with a communication aid, SS and TIS.

Ventilation

Two users required wheelchair structures to support their oxygen cylinders. One was a 17-year-old male with Morquio's disease complicated by lumbar and cervical spine fusions, hip and knee surgery and residual severe pain. He was also prescribed SS and TIS. The other was a 59-year-old male with motor neurone disease who also needed assessment for an environmental control unit. He was also provided with TIS but did not require SS.

Discussion

The 54 EPIOC users with RDs reported in this paper are a heterogeneous group, many with conditions rarely seen in clinical practise. Nonetheless, they make up 10% of the whole EPIOC cohort.[32] This is the first study of EPIOC users with RDs that focuses on the implications for the wheelchair components of rehabilitation. This may reflect the emphasis placed historically on research into the genetics and diagnosis of these RDs and the previously low level of support for younger physically disabled individuals in the UK.[62,63] However, these individuals with RDs will seldom be seen in locality-based rehabilitation services. It is important that the proposed centres for the study of these conditions [8,9] include rehabilitation expertise.

For those with inherited conditions, the progress of each individual is unique depending on activity levels, growth rate and development. The challenge for EPIOC providers is to reconcile the physical disabilities with the individual's need for function and participation whilst allowing for future disease progression and/or growth (for children). This is particularly important for those with small stature, for example, Morquio's disease and for those with extreme vulnerability, for example, osteogenesis imperfecta. This is illustrated by the individual with Morquio's disease who needed a complex prescription to accommodate the sequelae of his multiple orthopaedic surgery and need for oxygen. The SS (bespoke-carved foam) supported his joint derangements, while the TIS helped to minimise his problematic pain. He was provided with a 6-wheeled EPIOC providing a more stable base for a chair needing to accommodate an oxygen cylinder. However, for this individual, his residual abilities enabled him to control his chair using a standard joystick.

Table 1. Diagnosis, incidence/prevalence and effects of rare diseases in 54 electric powered indoor/outdoor wheelchair users.

Condition	Also called	Incidence/prevalence*	Effects
Achondroplasia	Achondroplastic dwarfism	1:26 000–34 608 [38]	Mutation of fibroblast growth factor receptor
Arthrogryposis	ARC Syndrome	1:3000 [39]	Soft-tissue, joint & skeletal deformity
Ataxia telangectasia		0.4:100 000* [40]	Progressive difficulty with coordinating movements (ataxia)
Central core disease	Shy–Magee syndrome	<6:100 000 live births [41,42]	Congenital myopathy
Cerebellar syndromes		0.3–2:100 000 for spinocerebellar [43]	Dysfunction of balance and movement
Congenital myasthenia	Erb-Goldflam syndrome	Unknown–very rare [44]	Neuromuscular weakness
Dejerine–Sottas disease	HMSN TYPE 111	<1:1 000 000 [45]	Polyneuropathy
Dystrophia epidermolysis bullosa		12–19/million births [46,47]	Skin erosion and blistering
Dystrophia myotonica		10.6:100 000* [42]	Progressive muscle wasting and weakness
Familial spastic paraplegia	Hereditary spastic paraplegia Strümpell-Lorrain syndrome	1.5–2.7:100 000 [48]	Progressive and severe lower extremity weakness and spasticity.
Fibrodysplasia ossificans progressiva	Myositis ossificans	1:2 000 000* [49]	Ossification of connective tissue
Friedreich's ataxia		0.15:100 000* [40]	Dysfunction of balance, movement and proprioception
Guillain–Barre syndrome		0.34 and 1.34/100 000 [50]	Acute progressive muscle weakness
Infantile systemic hyalinosis	Hyaline fibromatosis syndrome	<1:1 000 000 (52 reported cases worldwide) [51]	Hyalin deposits in tissues
Keratoderma	Focal palmoplantar keratoderma	Unclear	Severe blisters and calluses on the feet
Krabbe's disease	Galactocerebrosidase deficiency; globoid-cell leukodystrophy	1:100 000* [52]	Cerebral demyelination
Leukodystrophy: undiagnosed		<1:7663 births [53]	Progressive demyelination resulting in widespread motor and sensory dysfunction
McCune–Albright syndrome	Polyostotic fibrous dysplasia	1:100 000–1 000 000 people worldwide [54]	Fibrous dysplasia of bone, progressive scoliosis, short stature
Morquio's disease	Mucopolysaccharidosis	1:100 000 births [55]	Enzyme deficiency
Motor neurone disease	Amyotrophic lateral sclerosis	0.6–2.4:100 000 [56]	Degeneration of motor neurones resulting in muscle weakness and wasting
Multisystem atrophy		0.6 cases per 100.000 [57]	Combination of parkinsonian, autonomic, cerebellar or pyramidal symptoms and signs
Osteogenesis imperfecta	Brittle bone disease	1:20 000 births [58]	Connective tissue
Pelizaeus–Merzbacher disease	Cockayne–Pelizaeus–Merzbacher disease; PMD	<1:100 000 [53]	Growth of the myelin sheath
Progressive supranuclear palsy		1 per 100 000 [59]	Severe parkinsonism
Sandhoff's disease	Sandhoff–Jatzkewitz–Pilz disease; Total hexosaminidase deficiency	1:422 000 [60]	Neuronal destruction in brain and spinal cord
Spondylocostal dysplasia	Jarcho–Levin syndrome; spondylocostal dysostosis;	0.25/10 000 births [39]	Severe malformations of the vertebral column and ribs
Winchester syndrome	Winchester disease	<1:1000 000 (10 patients reported up to 2001)* [61]	Short stature, generalised osteolysis and progressive painful arthropathy

*Prevalence.

Comorbidity and additional clinical features

The results show that only 30% of this cohort had a single diagnosis, the remainder presented with complexities including a range of comorbidities and ACFs. It is often difficult to determine whether clinical issues are due to the condition itself or the physical problems caused by disability and immobility. Therefore, while epilepsy is a noted comorbidity in familial spastic paraplegia, it is a known ACF of Pelizaeus–Merzbacher disease. Epilepsy is not a contraindication to EPIOC use providing the user is daytime grand mal fit-free for at least one year, similar to the implications for drivers of motor vehicles.[64]

It is thought that individuals with Friedreich's ataxia are predisposed to developing diabetes.[65] One user with Friedreich's ataxia had diabetes which was recognised as an ACF. There may be no immediate implications for EPIOC prescription in those with uncomplicated diabetes, although it may eventually predispose users to pressure sores and leg ulcers. In contrast, one user with motor neurone disease also had diabetes complicated with a below-knee amputation which was noted as a comorbidity. For those with severe immobility disability, such as to require an EPIOC, dietary advice seems critical to prevent weight gain, obesity and minimise diabetic risk (as noted with multiple sclerosis [21]).

Pain was a common clinical finding in this group with problematic pain affecting 10 users. Provision of a wheelchair in individuals with Morquio's disease has been reported to alleviate pain and reduce fatigue, although it is also indicated that health-related QoL is reduced in wheelchair users.[19] It is likely that a similar situation applies to many EPIOC users and the provision

Table 2. Clinical and demographic features or 54 EPIOC users and their wheelchair provision.

Condition	No (male)	Comorbidities (cases)	Additional clinical features (cases)	Main diagnosis only	SS	TIS	Mean age (range)
Achondroplasia[b,c]	2 (0)	SCI (1): OA, DB, hypertension (1)	Pressure sore (1)	0	1	0	62.5 (60–65)
Arthrogryposis[b,c]	4 (1)	Skin rash (1): OA + BP and NP (1)	Painful post-hip replacement (1): scoliosis + contractures (1): OA hips (1)	1	0	4	31.5 (15–49)
Ataxia telangectasia[c]	1 (1)			1	0	1	44
Central core disease[c]	1 (1)			1	0	1	19
Cerebellar syndromes (2 cerebellar ataxia) (2 spino-cerebellar ataxia[c])	4 (0)		Problematic spasticity (1)	3	0	3	49.8 (28–61)
Congenital myasthenia[c]	1 (1)			1	0	1	11
Dejerine-Sottas disease[c]	1 (1)		Deep vein thrombosis and pulmonary embolism (1)	0	0	0	43
Dystrophia epidermolysis bullosa[b,c]	1 (1)		Abdominal pain? related to skin (1)	0	0	0	15
Dystrophia myotonica[c]	1 (1)	Problematic painful BP/coccidynia (1)	Swallowing difficulty (1)	0	0	0	54
Familial spastic paraplegia[c]	2 (2)	Epilepsy + hypertension + BP + ankle pain (1)		1	1	0	45.5 (23–68)
Fibrodysplasia ossificans progressiva[b,c]	1 (1)	BP (1)	Scoliosis with pelvic obliquity + problematic pain (1)	0	1	1	23
Friedreich's ataxia[c]	10 (5)	Psoriasis (1): BP + hypertension (1): BP (1): colonic + nasal polyps + peptic ulcer + BP (1)	Scoliosis (2): DB (1): choking/swallowing difficulties (1): aortic valve disease (1): obesity + oedema + problematic pain (1): oedema (1): scoliosis + problematic pain + choking (1): hypertrophic cardiomyopathy + pressure sore (1): problematic pain + oedema (1)	1	4	7	29.1 (16–43)
Guillain-Barre syndrome	1 (0)	OA knees and hands with failed surgery + asthma + hypertension (1)		0	0	1	68
Infantile systemic hyalinosis[b,c]	2 (0) sisters		Scoliosis (1): scoliosis + fragile skin (1)	0	2	2	15 (14–16)
Keratoderma[b,c]	1 (1)	Hypermobility (1)	Severe pain (1)	0	0	0	23
Krabbe's disease[c]	1 (0)		Communication impairment (1)	0	1	1	20
Leukodystrophy: undiagnosed[c]	1 (0)			1	1	0	13
McCune–Albright syndrome[b]	1 (0)		Multiple fractures + precocious puberty + Cushing's syndrome (1)	0	1	1	15
Morquio's disease[b,c]	2 (1)	Asthma (1)	Severe pain following spinal fusions + ventilatory failure (1): Previous two spinal fusions (1)	0	1	2	23 (17–29)
Motor neurone disease	6(5)	DB + below knee amputation (1)	Ventilatory failure (1)	4	0	4[a]	58 (51–63)
Multisystem atrophy	1 (0)		Postural hypotension (1)	0	0	0	58
Osteogenesis imperfecta[b,c]	4 (2)	Asthma (2)	Painful scoliosis + impaired hearing (1)	1	3	3	35 (17–60)
Pelizaeus–Merzbacher disease[c]	1 (1)		Epilepsy (1)	0	1	0	36
Progressive supranuclear palsy	1 (0)	Hypertension + irritable bowel syndrome + diverticular disease (1)		0	0	0	70
Sandhoff's disease[c]	1 (1)	Shoulder pain (wheelchair user's) (1)		0	0	1	52
Spondylocostal dysplasia[b,c]	1 (1)			1	0	0	19
Winchester syndrome[b,c]	1 (0)		Polyarthralgia + NP + oedema (1)	0	0	1	34
Total	54 (27)	35 (19)	45 (31)	16	17	34[a]	37.3 (11–70)

DB: Diabetes, OA: osteoarthritis, SCI: spinal cord injury, BP: back pain, NP: neck pain.
[a]TIS unknown for one user.
[b]Disorders involving connective tissues.
[c]Autosomal dominant, recessive or X-linked inheritance.

of TIS is one strategy for alleviating this pain.[66,67] In this group with RD, there is a preponderance, not seen in other studies, of powered wheelchair users with diseases affecting the musculoskeletal system, including the need for orthopaedic surgery with risks of post-surgical pain, which may be alleviated by SS and TIS. Some ACFs noted are those that would be associated with prolonged sitting in a wheelchair including pressure sores, oedema and thromboembolism.

Wheelchair features

The eight users with clinically significant scoliosis present specific challenges for EPIOC providers. An appropriate balance must be sought between stabilising the spine and retaining flexibility in the wheelchair system to maximise residual upper limb and trunk function. This was resolved by providing TIS for flexibility and pain management to all eight users with scoliosis and SS to the six users needing extra support. While surgery can ameliorate the progression of a scoliosis,[68] for many a scoliosis needs postural support by using SS to maintain posture and thus improve function.[16] The significant finding that those provided with SS was younger that those without such provision is likely to reflect the need for postural stability especially during growth.

Our largest group were those with Friedreich's ataxia ($n = 10$). It is recognised that, although some users with Friedreich's ataxia become unable to control their wheelchair,[18] many remain able to do so without use of non-standard control systems or use of head or foot controls as shown in this study. This possibly reflects the fact that weakness is not the primary impairment for those with Friedreich's ataxia.[69] Problematic pain was an issue for many Friedreich's ataxia users and seven of the users had TIS which would help to manage pain.[70,71]

Complex controls are needed when the user cannot manage a standard joystick. For those with substantial upper limb weakness and residual manual dexterity, the use of tray-mounted controls provide support for the weak upper limb allowing movement of the hand and fingers to be utilised. This was the case for two users, one with osteogenesis imperfecta and the other with familial spastic paraplegia. Tray-mounted controls also facilitate interfacing controls for those who need additional electronic assistive technologies, as in the case of the user with osteogenesis imperfecta. For one user (with infantile systemic hyalinosis) with extremely limited manual dexterity, the option of extra sensitive controls enabled her to remain in control of her chair. Tray-mounted joysticks may compete with space needed (e.g. for computers).

Rehabilitation issues

This paper contributes to the care pathways and clinical competencies that the UK Department of Health is striving to achieve.[6] Although rehabilitation is traditionally considered to be assisting recovery, rehabilitation professionals should also facilitate community living and participation for those with deteriorating conditions, which may be very hard to live with.[18] Often this will require assistive technologies including powered mobility being provided [14] and is best effected by a comprehensive service delivered by a multiprofessional team [72] including rehabilitation engineers skilled in assistive technology (as provided for our users). Previous research has shown that users and their families are generally satisfied with the EPIOC service provided,[30,73] but some were concerned that they would not be assessed for their changing needs as they had deteriorating conditions. [74] This is particularly important for those with RD, many of whom will deteriorate over time.

For those with inherited RD, other family members may have developed an identical or similar disease. This was demonstrated by the two sisters with infantile systemic hyalinosis who needed a high level of family support and when provided with EPIOCs, required a larger home which the rehabilitation team recommended.

Although it is reported that health related QoL is reduced and carer burden increased in wheelchair users with Morquio's disease,[19] there is good evidence that provision of an EPIOC improves quality of life [24] and reduces caregiver burden, particularly as the need to push a manual wheelchair is reduced.[74]

Although 75% of RD are in children,[75] some conditions may not have progressed to severe mobility disability until the individuals have reached adulthood. In our cohort, two such examples are EPIOC users with Sandhoff's disease and Pelizaeus–Merzbacher disease who were aged 52 and 36, respectively. What is unclear from data we were able to obtain was information about their rehabilitation pathway that led them to referral for an EPIOC (noting that those in the United Kingdom could self-refer to a wheelchair service). However, recent European recommendations for the management of mucopolysaccharidosis type 11 focuses on multidisciplinary team support, including physiotherapy to maintain ambulation with assistive devices if needed.[76] The lack of any mention of assistive technology in that review is not atypical. It reflects the lack of understanding of powered mobility as a major therapeutic tool,[77] enhancing mood through greater independence and social interaction, reducing pain, assisting swallowing and ventilation on occasions, and reducing caregiver burden.

Study limitations

It is recognised that the ACFs of these rare diseases may be incomplete or imprecise due to the paucity of literature reporting long-term follow-up of these individuals.[7] However, in the future, the development of registries for RDs will improve clinical data collection.[7] A combination of the rarity of the disease and progression to severe mobility disability resulted in a modest sized group for this study. Currently, there is a lack of evidence to indicate what proportions of those with RDs will progress to requiring an EPIOC.

Because data were extracted from records that were designed for clinical use, only data relevant to EPIOC prescription were recorded. The data represent the clinical picture at a particular time, often when their condition is deteriorating which creates specific issues for wheelchair services.[67] This may limit generalisability to other powered wheelchair populations, although the majority of this RD group are likely to progress. Service reorganisation prevented further follow-up of these users.

Our study did not include those who had purchased wheelchairs privately or through charitable funding (more often available for children). Users of mobility scooters were not included.

Conclusions

These EPIOC users with rare diseases reached the wheelchair service in their adult or teenaged years despite having an inherited and incurable (and often progressive) health condition. Their complex and diverse clinical problems presented unique challenges to the multi-professional wheelchair team to maintain successful community living. Combinations of problems arising from the RD trajectory, complications of disability and the acquisition of comorbidities presents a complex clinical picture that may appear daunting to rehabilitation professionals. This is compounded by a lack of research data on the rehabilitation of those with RDs. Our research has demonstrated that a multiprofessional rehabilitation team skilled in mobility assistive technology can resolve these challenges by approaching powered wheelchair provision from a therapeutic perspective to achieve independent mobility.

The recommended national strategies for RD [6,78,79] need to include rehabilitation in all its complexity and the potential of assistive technology to improve the well-being of those with RD and their families. Early assessment and regular review may help to address problems of severe disability and clinical complications before they have become established and require complex remedial, rehabilitation and medical interventions.

Declaration of interest

The authors declared that they have no conflicts of interest.

References

1. Health & Consumer Protection Directorate – General EC. Useful information on rare diseases from an EU perspective. Luxembourg: European Commission; 2014.
2. Genetic and Rare Diseases Information Center (GARD). Gaithersburg, MD 20898-8126, HHS.gov: U.S. Department of Health & Human Services; 2014; [cited 2015 August 3]. Available from: https://rarediseases.info.nih.gov/research/pages/41/rare-diseases-clinical-research-network.
3. National Institutes of Health Rare Diseases. US National Library of Medicine, 2015; [Cited 2015 August 3]. Available from: http://www.nlm.nih.gov/medlineplus/rarediseases.html.
4. Orphanet Report Series: Rare Diseases Collection Prevalence and incidence of rare diseases: bibliographic data. Diseases listed by decreasing prevalence, incidence or number of published cases. [cited 2015 August 3]. Available from: http://www.orpha.net/orphacom/cahiers/docs/GB/Prevalence_of_rare_diseases_by_decreasing_prevalence_or_cases.pdf.
5. Anonymous. Rare Disease Impact Report: insights from patients and the medical community. Shire Human Genetic Therapies, 2013: 1–34; [cited 2015 August 3]. Available from: http://www.geneticalliance.org.uk/docs/e-update/rare-disease-impact-report.pdf.
6. Department of Health. The UK strategy for rare diseases. London: Department of Health; 2013.
7. Rare Diseases UK. Improving lives, optimising resources: a vision for the UK rare disease strategy. London: Rare Disease UK; 2011.
8. Rare Disease UK. Centres of excellence for rare diseases. London: Rare Disease UK; 2013.
9. Rare Disease UK. Rare disease care coordination: delivering value, improving services. London: Rare Disease UK; 2013.
10. Rare Disease UK. Europlan: a national conference report. London: Rare Disease UK; 2014.
11. Rare Disease UK. Patients experiences of transition between care providers. London: Rare Disease UK; 2014.
12. Baumer D, Talbot K, Turner MR. Advances in motor neurone disease. J R Soc Med. 2014;107:14–21.
13. Gerstenecker A, Duff K, Mast B, et al. Behavioral abnormalities in progressive supranuclear palsy. Psychiatry Res. 2013;210:1205–1210.
14. Frank AO. Motor neurone disease: practical update ignores rehabilitative approaches – particularly assistive technology. Clin Med. 2010;10:640–641.
15. Rolfe J. Planning wheelchair service provision in motor neurone disease: implications for service delivery and commissioning. Brit J Occup Ther. 2012;75:217–222.
16. Clark J, Michael S, Morrow M. Wheelchair postural support for young people with progressive neuromuscular disorders. Int J Ther Rehabil. 2004;11:3765–3373.
17. Levy C, Berner TF, Sandhu PS, et al. Mobility challenges and solutions for fibrodysplasia ossificans progressiva. Arch Phys Med Rehabil. 1999;80:1349–1353.
18. Gibilisco P, Vogel AP. Friedreich ataxia. BMJ (Online). 2013;347:f7062.

19. Hendriksz CJ, Lavery C, Coker M, et al. Burden of disease in patients with Morquio A syndrome: results from an international patient-reported outcomes survey. Orphanet J Rare Dis. 2014;9:32. doi: 10.1186/1750-1172-9-32 20.
20. Hilbert JE, Kissel JT, Luebbe EA, et al. If you build a rare disease registry, will they enroll and will they use it? Methods and data from the national registry of myotonic dystrophy (DM) and facioscapulohumeral muscular dystrophy (FSHD). Contemp Clin Trials. 2012;33:302–311.
21. De Souza LH, Frank AO. Problematic clinical features of powered wheelchair users with severely disabling multiple sclerosis. Disabil Rehabil. 2015;37:990–996.
22. Frank AO, Ward JH, Orwell NJ, et al. Introduction of a new NHS electric-powered indoor/outdoor chair (EPIOC) service: benefits, risks and implications for prescribers. Clin Rehabil. 2000;14:665–673.
23. Frank AO, Ellis K, Yates M. Use of the voucher scheme for provision of electric powered indoor/outdoor wheelchairs (EPIOCs). ''Posture & Mobility'' 2008;23:17–26.
24. Davies A, De Souza LH, Frank AO. Changes in the quality of life in severely disabled people following provision of powered indoor/outdoor chairs. Disabil Rehabil. 2003;25:286–290.
25. De Michele G, Perrone F, Filla A, et al. Age of onset, sex, and cardiomyopathy as predictors of disability and survival in Friedreich's disease: a retrospective study on 119 patients. Neurol. 1996;47:1260–1264.
26. Martin J, Martin L, Lofgren A, et al. Classical Friedreich's ataxia and its genotype. Eur Neurol. 1999;42:109–115.
27. Muller-Felber W, Rossmanith T, Spes C, et al. The clinical spectrum of Friedreich's ataxia in German families showing linkage to the FRDA locus on chromosome 9. Clin Invest. 1993;71:109–114.
28. Klockgether T, Ludtke R, Kramer B, et al. The natural history of degenerative ataxia: a retrospective study in 466 patients. Brain. 1998;121:589–600.
29. Schoser B, Sommer C, Dingermann T. Morbus pompe and morbus fabry: rare treatable metabolic diseases [German] Morbus Pompe und Morbus Fabry: seltene therapierbare stoffwechselerkrankungen mit bedeutung fur den neurologen. Psychopharmakotherap. 2009;16:192–197.
30. Evans S, Neophytou C, De Souza LH, et al. Young people's experiences using electric powered indoor-outdoor wheelchairs (EPIOCs): potential for enhancing users' development? Disabil Rehabil. 2007;19:1281–1294.
31. Axelson P, Zollars JA. Presentation on assistive technologies for the seating and mobility needs of persons with osteogenesis imperfecta. Connect. Tissue Res. 1995;31:S45–S47.
32. Frank AO, De Souza LH. Recipients of electric-powered indoor/outdoor wheelchairs provided by a national health service: a cross-sectional study. Arch Phys Med Rehabil. 2013;94:2403–2409.
33. Bostrom K, Ahlstrom G. Living with a chronic deteriorating disease: the trajectory with muscular dystrophy over ten years. Disabil Rehabil. 2005;26:1388–1398.
34. Naidoo V, Putnam M, Spindel A. Key focal areas for bridging the fields of aging and disability: findings from the growing older with a disability conference. Int J Integr Care. 2012;12:e201
35. Mangin D, Heath I, Jamoulle M. Beyond diagnosis: rising to the multimorbidity challenge. BMJ (Online). 2012;344: 7865. (doi:http://dx doi org/10 1136/bmj e3526 344.
36. Greer N, Brasure M, Wilt TJ. Wheeled mobility (wheelchair) service delivery: scope of the evidence. Ann Intern Med. 2012;156:141–146.
37. British Society of Rehabilitation Medicine. Specialised wheelchair seating national clinical guidelines. Report of a multidisciplinary expert group (Chair: Marks LJ). London: British Society of Rehabilitation Medicine; 2004.
38. Barbosa-Buck CO, Orioli IM, da M, et al. Clinical epidemiology of skeletal dysplasias in South America. Am J Med Genet A. 2012;158A:1038–1045.
39. Campbell RM. Jr., Spine deformities in rare congenital syndromes: clinical issues. Spine. 2009;34:1815–1827.
40. Erichsen A, Koht J, Stray-Pedersen A, et al. Prevalence of hereditary ataxia and spastic paraplegia in southeast Norway: A population-based study. Brain. 2009;132:1577–1588.
41. Jungbluth H. Central core disease. Orphanet J Rare Dis. 2007;25. doi:10.1186/1750-117 2-2.
42. Norwood FL, Harling C, Chinnery PF, et al. Prevalence of genetic muscle disease in Northern England: in-depth analysis of a muscle clinic population. Brain. 2009;132:3175–3186.
43. Van De Warrenburg BPC, Sinke RJ, Verschuuren-Bemelmans CC, et al. Spinocerebellar ataxias in the Netherlands: prevalence and age at onset variance analysis. Neurol. 2002;58:702–708.
44. Pitt M. Neurophysiological strategies for the diagnosis of disorders of the neuromuscular junction in children. Dev Med Child Neurol. 2008;50:328–334.
45. Dutch Neuromuscular Research Centre. Dejerine Sottas Syndrome. the Netherlands, Dutch Neuromuscular Research Centre. Neuromuscular information, disorders and diagnostics. 2003.
46. Browne F, Heagerty AHM, Martinez A, et al. The epidemiology of epidermolysis bullosa in the U.K: a 9-year study. Brit J Dermatol. 2011;165:0007–0963.
47. Kho YC, Rhodes LM, Robertson SJ, et al. Epidemiology of epidermolysis bullosa in the antipodes: the Australasian epidermolysis bullosa registry with a focus on Herlitz junctional epidermolysis bullosa. Arch Dermatol. 2010;146:635–640.
48. Fortini D, Cricchi F, Di Fabio R, et al. Current insights into familial spastic paraparesis: new advances in an old disease. Funct Neurol. 2003;18:43–49.
49. Miao J, Zhang C, Wu S, et al. Genetic abnormalities in fibrodysplasia ossificans progressiva. Genes Genet Syst. 2012;87:213–219.
50. McGrogan A, Madle GC, Seaman HE, et al. The epidemiology of Guillain-Barré syndrome worldwide. A systematic literature review. Neuroepidemiology. 2009;32:150–163.
51. Lindvall LE, Kormeili T, Chen E, et al. Infantile systemic hyalinosis: case report and review of the literature. J Am Acad Dermatol. 2008;58:303–307.
52. Deane JE, Graham SC, Kim NN, et al. Insights into Krabbe disease from structures of galactocerebrosidase. Proc Natl Acad Sci USA. 2011;108:15169–15173.

53. Bonkowsky JL, Nelson C, Kingston JL, et al. The burden of inherited leukodystrophies in children. Neurology. 2010;75:718–725.
54. Dumitrescu CE, Collins MT. McCune-Albright syndrome. Orphanet J Rare Dis. 2008;3. doi: 10.1186/1750-1172-3-12
55. Matallana AM, De LM, Palma MA, et al. Morquio disease type IVA: clinical cases. Hormone Res Paed. 2012;78: 1663–2818.
56. Cronin S, Hardiman O, Traynor BJ. Ethnic variation in the incidence of ALS: a systematic review. Neurology. 2007;68:1002–1007.
57. Vanacore N. Epidemiological evidence on multiple system atrophy. J Neural Transm. 2005;112:1605–1612.
58. Engelbert RH, Pruijs HE, Beemer FA, et al. Osteogenesis imperfecta in childhood: treatment strategies. Arch Phys Med Rehabil. 1998;79:1590–1594.
59. Golbe LI. The epidemiology of progressive supranuclear palsy. The Netherlands:Elsevier; 2008.
60. Meikle PJ, Hopwood JJ, Clague AE, et al. Prevalence of lysosomal storage disorders. JAMA. 1999;281:249–254.
61. Matthiesen G, Faurholt V, Helin P, et al. Winchester syndrome. Internat Orth. 2001;25:331–333.
62. Clarke S, Sloper P, Moran N, et al. Multi-agency transition services; greater collaboration needed to meet the priorities of young disabled people with complex needs as they move into adulthood. J Integrated Care. 2011;19:30–40.
63. Knight KH, Porcellato L, Tume L. Out-of-school lives of physically disabled children and young people in the United Kingdom: a qualitative literature review. J Child Health Care. 2014;18:275–285.
64. Driver and Vehicle Licensing Authority. Driving licences – epilepsy: Group 1 (car + motorcycle) driving entitlement. IN59. 2014. Available from: https://www.gov.uk/government/uploads/system/uploads/attachment_data/file/380806/INS9.pdf on 3rd August 2015
65. Parkinson MH, Boesch S, Nachbauer W, et al. Clinical features of Friedreich's ataxia: classical and atypical phenotypes. J Neurochem. 2013;126:103–117.
66. Frank AO, De Souza LH, Frank JL, et al. The pain experiences of powered wheelchair users. Disabil Rehabil. 2012;34:770–778.
67. Richardson M, Frank AO. Electric powered wheelchairs for those with muscular dystrophy: problems of posture, pain and deformity. Disabil Rehabil Assist Technol. 2009;4: 181–188.
68. Milbrandt TA, Kunes JR, Karol LA. Friedreich's ataxia and scoliosis: The experience at two institutions. J Pediatr Orthop. 2008;28:234–238.
69. Beauchamp M, Labelle H, Duhaime M, et al. Natural history of muscle weakness in Friedreich's Ataxia and its relation to loss of ambulation. Clin Orth Rel Res. 1995;311:270–275.
70. Ding D, Leister E, Cooper RA, et al. Usage of tilt-in-space, recline, and elevation seating functions in natural environment of wheelchair users. J Rehabil Res Dev. 2008;45:973–984.
71. Lacoste M, Weiss L, Allard M, et al. Powered tilt/recline systems: why and how are they used? Assist Technol. 2003;15:58–68.
72. Leigh PN, Abrahams S, Al-Chalabi A, et al. The management of motor neurone disease. J Neurol Neurosurg Psychiat. 2003;74:iv32–iv47.
73. Evans S, Frank A, Neophytou C, et al. Older adults' use of, and satisfaction with, electric powered indoor/outdoor wheelchairs. Age Ageing 2007;36:431–435.
74. Frank AO, Neophytou C, Frank J, et al. Electric powered indoor/outdoor wheelchairs (EPIOCs): users views of influence on family, friends and carers. Disabil Rehabil Assist Technol. 2010;5:327–338.
75. National Alliance for People with Rare Diseases and all who support them. About rare diseases. Available from http://www.raredisease.org.uk/on. [Cited 2015 August 3].
76. Scarpa M, Almássy Z, Beck M, et al. Mucopolysaccharidosis type II: European recommendations for the diagnosis and multidisciplinary management of a rare disease. Orphanet J Rare Dis. 2011;6:72. doi:10.1186/1750-1172-6-72
77. Frank AO, De Souza LH. Therapeutic roles of powered indoor-outdoor wheelchairs in multiple sclerosis. Equipment Serv. 2015;82–85.
78. Kodama TK. Global strategy for rare and intractable diseases. Clin Neurol. 2013;53:1283–1286.
79. Taruscio D, Gentile AE, De Santis M, et al. EUROPLAN: a project to support the development of national plans on rare diseases in Europe. Public Health Genomics. 2013;16:278–287.