

Rare diseases and powered mobility

Rare diseases: matching wheelchair users with rare metabolic, neuromuscular or neurological disorders to Electric Powered Indoor/Outdoor Wheelchairs (EPIOCs).

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⁺ Stanmore Specialist Wheelchair Service has now been disbanded.

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Abstract

Introduction: Little is written about the rehabilitation of those with rare diseases and their use of powered wheelchairs.

Purpose: The aim of this study is to describe the clinical features of electric powered indoor/outdoor wheelchairs (EPIOC) users with rare diseases (RD) that impact on EPIOC provision and seating needs.

Method: Retrospective review of electronic and case note records of EPIOC recipients attending a specialist wheelchair service between June 2007 and September 2008.

Records were reviewed by a consultant in rehabilitation medicine, data systematically extracted and entered into a computer database. Further data were entered from clinical records and extracted under three themes; demographic, diagnostic, clinical (including comorbidity and associated features of the illness/disability (ACFs)) and wheelchair factors. Records were reviewed a mean of 64 (range 0-131) months after receiving their wheelchair.

Results: Fifty four (27 male) EPIOC users, mean age 37.3 (sd 18.6, range 11-70) met the inclusion criteria. Diagnoses included Friedreich's ataxia (n=10), motorneurone disease (n=6), osteogenesis imperfecta (n=4), arthrogyrosis (n=4), cerebellar syndromes (n=4) and others (n=26). 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%). Those provided with SS were significantly younger than those who had standard equipment ($p<0.004$). Four users had between them complex control or interfacing issues. Two users required support for oxygen cylinders.

Conclusions: This study contributes to the limited understanding of the rehabilitation needs of severely mobility impaired individuals with rare diseases from the perspective of a powered

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wheelchair service. Rehabilitation is complicated by comorbidity and the complex clinical findings in this group of wheelchair users.

Key words: comorbidity, Friedreich's ataxia, rare diseases, rehabilitation, wheelchairs

Rare diseases (RD) are conditions affecting less than five in 10,000 of the general population¹, or a prevalence of fewer than 200,000 affected individuals in the USA². Many are life-threatening or chronically debilitating diseases, often of genetic origin¹. Whilst the diagnosis and medical management of RD have advanced, little is published on the functional impact on individuals with these disorders experiencing progressive disabilities.

It has been recommended that more research is needed into management of patients with RD to underpin the development of guidelines to improve care³.

For those severely affected by RD, mobility disability has an impact on activities, participation and quality of life (QoL). The consideration of wheeled and/or powered mobility can provide substantial improvements to QOL⁴. The limited literature on rehabilitation and RD, and the even rarer literature on wheelchair use, appears negative in nature – in that a condition had deteriorated such that a wheelchair was needed for mobility⁵⁻⁹. UK Reports make no reference to mobility disability and the need for a full disability assessment and rehabilitation³. This is surprising in view of the proven benefits of powered mobility to the wellbeing of EPIOC users (referred to as ‘users’), particularly the psychological gains experienced by younger users¹⁰. However the value of assistive technologies for those with RD and their families is understood and promoted in the USA¹¹.

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 prescription¹². We explored a subgroup of this study population with severe mobility disability diagnosed with RDs that were of metabolic, neuromuscular or neurological origin.

The aim of this study is to describe those clinical features of EPIOC users with RD that impact on EPIOC provision and seating needs.

Methods

The setting

The Specialist Wheelchair Service at Stanmore was set up in 1997¹³ 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)¹³.

Provision involved: -

1. Completion of a screening questionnaire.
2. Occupational therapy assessment for the suitability of the home environment and the likelihood that the eligibility criteria would be fulfilled.
3. Assessment at the 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.
4. 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 regional service. All individuals who had been prescribed an EPIOC and were currently using their chair were of interest to this study. Users relevant to this research had a diagnosis of a rare disease defined as conditions affecting less than five in 10,000 of the

general population¹, of metabolic, neurological or neuromuscular origin, and recorded as the main diagnosis for ten or fewer individuals. Data had been recorded by health professionals after physical examination. Assessment included demographic data, clinical issues and wheelchair factors. Data were entered into electronic records and were reviewed between June 2007 and September 2008 by a consultant physician in rehabilitation medicine. Data were systematically extracted and entered into a computer database for analysis. Further data were entered from the clinical notes (charts) and all data anonymised.

Data relating to demographic profile, clinical profile and wheelchair factors were extracted for analysis. Demographic profiles consisted of information on age at initial assessment and gender. Clinical profiles included: primary diagnosis, comorbidities, other clinical features and complications relating to the disability.

Wheelchair factors included information about specialised seating (SS)(adaptive seating), 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’¹⁴. Other data included tilt-in-space (TIS), cushions and complex controls.

Methods of analysis

This was a sub-group analysis of EPIOC users from a cohort reported elsewhere¹².

Descriptive statistics were used to analyse demographic and wheelchair data. 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). Other clinical data included comorbidities (conditions with no known or unlikely association with the index diagnosis) and additional clinical features (ACFs) which were features of the condition and/or features of disability likely to influence the wheelchair prescription.

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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) met the inclusion criteria. This was 10% of the original cohort¹². There were 27 males with mean age of 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 have 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) and six had motor neurone disease (five men, one woman mean age 58, range 51-63.). 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 5 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.

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Wheelchair features.

TIS was provided to 34 (63%) users and SS to 17 (31%). Six users had individually tailored seating systems. Carved foam seating was provided to three (achondroplasia, Friedreich's ataxia and infantile systemic hyalinosis), 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. 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 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 practice. Nonetheless they make up 10% of the whole EPIOC cohort¹². 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 UK³⁹⁻⁴⁰. 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 conditions⁴¹⁻⁴² 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 e.g. Morquio's disease and for those with extreme vulnerability e.g. osteogenesis imperfecta. This is illustrated by the individual with Morquio's disease (see results) who needed a complex prescription to accommodate the sequelae of his multiple orthopaedic surgery and need for oxygen. The SS (bespoke carved

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foam) supported his joint derangements whilst 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.

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 if 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 vehicles⁴³.

It is thought that individuals with Friedreich's ataxia are predisposed to developing diabetes⁴⁴. 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 sclerosis⁴⁵).

Pain was a common clinical finding in this group with problematic pain affecting 10 users. Provision of a wheelchair in people 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 users⁴⁶. It is likely that a similar situation applies to many EPIOC users and the

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provision of TIS is one strategy for alleviating this pain⁴⁷⁻⁴⁸. 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. Whilst surgery can ameliorate the progression of a scoliosis⁴⁹, for many a scoliosis needs postural support by using SS to maintain posture and thus improve function⁵⁰.

Our largest group were those with Friedreich's ataxia (n=10) and it is recognised that although some users with Friedreich's ataxia become unable to control their wheelchair⁵¹, this study confirms that many remain able to do so without use of non-standard control systems or use of head or foot controls. This possibly reflects the fact that weakness is not the primary impairment for those with Friedreich's ataxia⁵². Problematic pain was an issue for many Friedreich's ataxia users and seven of the users had TIS which would help to manage pain⁵³⁻⁵⁴.

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 utilized. This was the case for two users, one with osteogenesis imperfecta and the other with familial spastic paraplegia. Tray-mounted joysticks are usually centrally positioned and compete with space needed (e.g. for computers). 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.

Rehabilitation issues

This paper contributes to the care pathways and clinical competencies that the UK Department of Health is striving to achieve⁵⁵. 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 with⁵¹. Often this will require assistive technologies including powered mobility being provided⁵⁶ and is best effected by a comprehensive service delivered by a multiprofessional team⁵⁷ including rehabilitation engineers skilled in assistive technology. The EPIOC service at Stanmore consisted of a multiprofessional team embracing a rehabilitation physician, therapists and engineers/clinical scientists able to provide a holistic approach to improving mobility and the facility to refer onwards where needed. Previous research has shown that users and their families are generally satisfied with the EPIOC service provided^{10,58}, but some were concerned that they would not be assessed for their changing needs as they had deteriorating conditions⁵⁸. This is particularly important for these RD, many of which will deteriorate over time.

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The inclusion of the needs of caregivers and families in the rehabilitation assessment is particularly important in EPIOC provision for those with inherited RD as 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 disease⁴⁶ there is good evidence that provision of EPIOC improves quality of life⁴ and reduces caregiver burden, particularly as the need to push a manual wheelchair is reduced⁵⁹.

Although 75% of RD are in children⁶⁰, 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 needed⁶¹. 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, enhancing mood through greater independence and social interaction, reducing pain, assisting swallowing and ventilation on occasions, and reducing caregiver burden.

Limitations of study

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. Consequently our categorisation was led by clinical judgement and the available history of each user. Because data were extracted from records that were designed for clinical use, only data relevant to EPIOC prescription were recorded. The data represents the clinical picture at a particular time, which may limit generalisability to other powered wheelchair populations. 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), nor scooters. Many of these users would have maintained balance ability and so those without severe deformity might have purchased scooters.

Conclusions

Our EPIOC users with rare diseases reached the wheelchair service in their adult or teenaged years despite having an inherited and incurable progressive health condition. Their complex and diverse clinical problems presented unique challenges to the multi-professional wheelchair team to maintain successful community living. Early intervention is recommended and should take a health promotion approach to the long-term management of rare diseases through the provision of EPIOCs and specialised seating. This is preferable to addressing problems of severe disability and clinical complications once they have become established and when complex remedial, rehabilitation and medical interventions are required.

The recommended national strategies for RD^{55,62-63} need to include rehabilitation in all its complexity and the potential of assistive technology to improve the wellbeing of those with RD and their families.

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 ¹⁵	Mutation of fibroblast growth factor receptor
Arthrogryposis	ARC Syndrome	1: 3,000 ¹⁶	Soft tissue, joint & skeletal deformity
Ataxia telangectasia		0.4:100,000 * ¹⁷	Progressive difficulty with coordinating movements (ataxia)
Central core disease	Shy-Magee syndrome	< 6: 100,000 live births ^{18;19}	Congenital myopathy
Cerebellar syndromes		0.3 – 2:100,000 for spinocerebellar ²⁰	Dysfunction of balance and movement
Congenital myasthenia	Erb-Goldflam syndrome	Unknown - very rare ²¹	Neuromuscular weakness
Dejerine-Sottas disease	HMSN TYPE 111	< 1: 1,000,000 ²²	Polyneuropathy
Dystrophia epidermolysis bullosa		12-19 / million births ^{23;24}	Skin erosion and blistering
Dystrophia myotonica		10.6: 100,000* ¹⁹	progressive muscle wasting and weakness
Familial spastic paraplegia	Hereditary spastic paraplegia	1.5 – 2.7:100,000 ²⁵	progressive and severe lower extremity weakness and spasticity.
Fibrodysplasia ossificans progressiva	Myositis Ossificans	1:2,000,000 * ²⁶	Ossification of connective tissue
Friedreich's ataxia		0.15:100,000 * ¹⁷	Dysfunction of balance, movement and proprioception
Guillain-Barre Syndrome		0.34 and 1.34/100,000 ²⁷	Acute progressive muscle weakness
Infantile systemic hyalinosis	Hyaline fibromatosis syndrome	< 1: 1,000,000 (52 reported cases world-wide) ²⁸	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* ²⁹	Cerebral demyelination
Leukodystrophy: undiagnosed		< 1 :7663 births ³⁰	Progressive demyelination resulting in widespread motor and sensory dysfunction
McCune-Albright syndrome	Polyostotic fibrous dysplasia	1: 100,000 - 1,000,000 people worldwide ³¹	Fibrous dysplasia of bone, progressive scoliosis, short stature
Morquio's disease	Mucopolysaccharidosis	1: 100,000 births ³²	Enzyme deficiency
Motor neurone disease	amyotrophic lateral sclerosis	0.6 – 2.4:100,000 ³³	Degeneration of motor neurones resulting in muscle weakness and wasting

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Multisystem atrophy		0.6 cases per 100.000 ³⁴	Combination of parkinsonian, autonomic, cerebellar or pyramidal symptoms and signs
Osteogenesis imperfecta	Brittle bone disease	1:20,000 births ³⁵	Connective tissue
Pelizaeus-Merzbacher Disease	Cockayne-Pelizaeus-Merzbacher Disease; PMD	<1:100,000 ³⁰	Growth of the myelin sheath
Progressive supranuclear palsy		1 per 100,000 ³⁶	Severe parkinsonism
Sandhoff's disease	Sandhoff-Jatzkewitz-Pilz disease; Total hexosaminidase deficiency	1:422,000 ³⁷	Neuronal destruction in brain and spinal cord
Spondylocostal dysplasia	Jarcho-Levin syndrome; spondylocostal dysostosis;	0.25 / 10,000 births ¹⁶	Severe malformations of the vertebral column and ribs
Winchester syndrome	Winchester Disease	<1 :1 000,000 (10 patients reported up to 2001)* ³⁸	Short stature, generalized osteolysis and progressive painful arthropathy

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†^	2 (0)	SCI (1): OA, DB, hypertension (1)	Pressure sore (1)	0	1	0	62.5 (60-65)
Arthrogyrosis†^	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^	1 (1)			1	0	1	44
Central core disease^	1 (1)			1	0	1	19
Cerebellar syndromes (2 cerebellar ataxia) (2 spino-cerebellar ataxia^)	4 (0)		Problematic spasticity (1)	3	0	3	49.8 (28-61)
Congenital myasthenia^	1 (1)			1	0	1	11
Dejerine-Sottas disease^	1 (1)		Deep vein thrombosis and pulmonary embolism (1)	0	0	0	43
Dystrophia epidermolysis bullosa†^	1 (1)		Abdominal pain? related to skin (1)	0	0	0	15
Dystrophia myotonica^	1 (1)	Problematic painful BP/coccydynia (1)	Swallowing difficulty (1)	0	0	0	54
Familial spastic paraplegia^	2 (2)	Epilepsy + hypertension + BP + ankle pain (1)		1	1	0	45.5 (23-68)
Fibrodysplasia ossificans progressiva†^	1 (1)	BP (1)	Scoliosis with pelvic obliquity + problematic pain (1)	0	1	1	23
Friedreich's ataxia^	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)

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Guillain-Barre syndrome	1 (0)	OA knees and hands with failed surgery +asthma + hypertension (1)		0	0	1	68
Infantile systemic hyalinosis ^{†^}	2 (0) sisters		Scoliosis (1): scoliosis + fragile skin (1)	0	2	2	15 (14-16)
Keratoderma ^{†^}	1 (1)	Hypermobility (1)	Severe pain (1)	0	0	0	23
Krabbe's disease [^]	1 (0)		Communication impairment (1)	0	1	1	20
Leukodystrophy: undiagnosed [^]	1 (0)			1	1	0	13
McCune-Albright syndrome [†]	1 (0)		Multiple fractures +precocious puberty + Cushing's syndrome (1)	0	1	1	15
Morquio's disease ^{†^}	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*	58 (51-63)
Multisystem atrophy	1 (0)		Postural hypotension (1)	0	0	0	58
Osteogenesis imperfecta ^{†^}	4 (2)	Asthma (2)	Painful scoliosis + impaired hearing (1)	1	3	3	35 (17-60)
Pelizaeus-Merzbacher disease [^]	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 [^]	1 (1)	Shoulder pain (wheelchair user's) (1)		0	0	1	52
Spondylocostal dysplasia ^{†^}	1 (1)			1	0	0	19
Winchester syndrome ^{†^}	1 (0)		Polyarthralgia + NP + oedema (1)	0	0	1	34
Total	54 (27)	35 (19)	45 (31)	16	17	34*	37.3 (11-70)

Key: † Disorders involving connective tissues.^ Autosomal dominant, recessive or X-linked inheritance. * TIS unknown for one user. DB: Diabetes, OA: osteoarthritis, SCI: spinal cord injury, BP: back pain, NP: neck pain

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