



## ORIGINAL RESEARCH ARTICLE

## Understanding Musculoskeletal Concerns of Adults with Congenital Upper Limb Differences: Prevalence, Impact on Function and Treatment

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### Abstract

Limited research exists regarding challenges encountered by individuals with congenital unilateral upper limb differences (CUULD). There is a gap in understanding the impact of these challenges on everyday activities and interventions received. This poses a challenge for rehabilitative professionals to tailor treatments to address the unique needs of this population. This study examined the musculoskeletal concerns of individuals with CUULD, the impact on daily activities and the satisfaction of treatments received for these concerns. Data was gathered from 95 participants with CUULD using a 19-question survey including symptoms, activities impacted, treatments received and perceived satisfaction of those interventions. Data was analyzed using descriptive statistics and frequency analysis. The results indicate that 82% of respondents have or currently experience musculoskeletal issues with 41% experiencing these by the age of 25. Neck pain was the highest reported symptom (51%) followed by general pain (43%) and tingling/numbness (42%). Treatments received indicate a wide variety with mixed satisfaction. Lastly, participants rated weightlifting/strength training as the most impacted activity with yard work/general maintenance rated second. This study highlights the impact of CUULD on individuals and their daily activities and underscores the need for interventions that target the symptoms experienced and address engagement in impacted activities.

### Keywords

Congenital unilateral upper limb difference, Rehabilitative professionals, Musculoskeletal, Treatment, Daily activities

### Abbreviations

ULD: Upper Limb Difference; CTS: Carpal Tunnel Syndrome; CUULD: Congenital Unilateral Upper Limb Difference

### Introduction

Individuals living with a difference to one or more upper limbs encounter a range of distinct challenges across their lifespan. Upper limb differences may result from acquired limb loss due to injury or medical conditions or a limb difference due to congenital abnormalities. For this study, the term upper limb difference (ULD) will represent both limb loss and limb difference.

### Incidence and prevalence of ULD

Estimates of incidence of ULD vary greatly, however two, 11-year long studies indicate the prevalence of congenital limb differences to be 19.76-21.5 out of 10,000 live births [1]. In 2005, the United States saw approximately 541,000 individuals living with upper limb loss, and an average of 1,500 individuals are born with congenital unilateral upper limb differences (CUULD) each year [2]. The most common types of limb difference seen are classified as failures of differentiation followed by duplications [1]. A retrospective analysis of 5 years of medical claims indicates acquired limb loss is considerably more common than congenital limb difference. However, individuals with congenital limb difference are found living in the general population at 50% more than those with acquired limb loss due to the often-seen comorbidities associated with limb loss [3]. This underscores the growing demand for rehabilitative services from multiple disciplines to support the

distinct needs of individuals with both limb loss and limb difference.

### **Pain and overuse**

Musculoskeletal pain and overuse injuries are common in both the affected and unaffected limbs for individuals with ULD. Upwards of 63% of individuals with congenital limb differences report experiencing chronic musculoskeletal pain [4]. Studies report individuals with limb difference experience overuse injuries in the contralateral limb including pain, carpal tunnel syndrome (CTS), tendonitis (shoulder, elbow, wrist), nerve damage and osteoarthritis with overall pain and CTS being the most prevalent [4-7]. One study showed that 26.6% of participants with congenital and acquired ULD reported CTS in the unaffected limb which is higher than the average population [8]. Pain is reported to be mostly in the no affected arm, back, and neck as well as the contralateral shoulder, wrist, and hand by individuals with limb differences [8,9].

This confirms that individuals living with a CUULD are more likely to experience overuse symptoms associated with pain that persist throughout life. Engaging in repetitive and/or resistive activities as well as the compensatory movements used by individuals with limb differences place them at higher risk of overuse syndrome [10,11]. Spinal deviations have been found to be present at greater rates than in those without limb differences which can contribute to the development of overuse and pain. Spinal deviations are likely due to the postural imbalances created by the asymmetries of the arms and trunk and use of compensatory movements [12,13]. Individuals with limb differences who experience additional orthopedic conditions demonstrate lower quality of life scores than even those with just a limb difference [14]. These studies indicate the potential impact of limb difference and associated musculoskeletal conditions on overall function and quality of life in individuals with limb difference.

### **Impact on function**

The musculoskeletal conditions associated with overuse and pain experienced by individuals with upper limb differences influence aspects of daily life such as participation and engagement in work [15]. Individuals living with ULD experience reduced physical and mental quality of life scores and greater disability scores when compared to the general population [11,13,15-17]. Adolescents have been shown to have fewer choices in a career due to being seen as less capable by educators and employers as well as requiring less physically demanding jobs [18]. According to Postema, et al. [9], 14.6% of individuals with ULD reported working less and 31% of individuals reported unemployment due to their upper limb difference. A study by Johansen, et al. [19] revealed that one third of participants retired early due to their congenital upper limb difference. Additionally,

27% percent of individuals with upper limb differences reported they had to adapt to their work environment due to their difference [19]. As work provides a sense of purpose and meaning for many individuals, interventions that allow individuals to maintain employment and engage in daily activities are necessary.

### **Existing treatment**

Limited literature was found specific to a variety of treatment options for CUULD. Surgical options may be recommended for many types of CUULD to attempt to allow for increased function and aesthetics. Options that allow for reconstruction of the impacted hand or arm have shown mixed outcomes however and lack consensus on treatment techniques [20]. Outside of surgical interventions, treatment for individuals with CUULD has traditionally included occupational therapy, physical therapy, and other medical interventions such as cortisone shots, nerve blockers, ultrasound, dry needling, and pain medication [21]. These varied treatments are used in hopes of relieving pain associated with overuse and speak to the involvement of multiple disciplines on the care of individuals with CUULD. Though not specific to the CUULD population, one study exploring the management of chronic and complex musculoskeletal pain highlighted the benefits of holistic, multidisciplinary teams in the care of patients who otherwise experience frustration with treatment options [22]. The frustrations noted by participants of that study speak to the difficulties faced in accessing effective treatments.

Individuals with CUULD encounter many barriers to receiving adequate care for their limb difference and associated musculoskeletal issues. Limited knowledge of congenital limb difference and a lack of care guidelines impacts access to care. In one study, participants felt that the lack of a therapeutic plan that addressed their personal needs was detrimental to their overall function. Additionally, limited knowledge of prosthetic options and other interventions creates barriers to accessing effective health care options [11]. Individuals with ULD may not be aware of the expertise of rehab professionals to address a full range of topics and often feel that periodic appointments are largely unhelpful and allow for fragmented care [22,23]. Lastly, individuals with ULD may lack knowledge regarding their own conditions and experience gaps in accessing rehabilitative services indicating a need for education for the limb different population as well as for the rehabilitation professionals who care for them [18,22,24].

### **Objectives**

The literature demonstrates that individuals with ULD present with musculoskeletal concerns of overuse and pain that may limit their ability to participate in meaningful activities. While this literature may be inclusive of those with CUULD, limited research

exists specific to that population. The aim of this study is to determine the musculoskeletal concerns of individuals with CUULD in depth. The research focused on the following questions. What are the musculoskeletal concerns experienced by individuals with CUULD? How do these concerns impact engagement in daily activities?

What interventions do individuals with CUULD receive for their musculoskeletal concerns and lastly, what is the satisfaction of these treatments? This information will be useful to inform approaches taken by rehabilitation specialists providing services to these individuals, providing a greater understanding of specific activities to address and treatment approaches to consider.

## Methods

### Research design

This quantitative, descriptive study employed a survey methodology to explore the musculoskeletal concerns associated with CUULD. The survey explored various aspects including musculoskeletal issues, treatments, interventions, and the utilization of adaptive devices among individuals with CUULD. Employing a purposive sampling strategy, the principal investigator distributed the survey through targeted social media groups comprised of members affected by CUULD. Leveraging contacts within the limb different community, the survey dissemination was further facilitated via social media platforms leading to a snowball sampling effect. Approval of the study was secured from the [redacted] Institutional Review Board, ensuring adherence to ethical standards. Prior to participation, participants were provided with informed consent.

### Participants

The study comprised a global sample of individuals with CUULD. Inclusion criteria stipulated participants to be 18 years or older and fluent in English, with congenital onset of their limb difference. Participants were recruited through social media groups specific to limb loss and limb difference via a post made by the author advertising the study between January 2024 and April 2024. This advertisement included a brief explanation of the study, eligibility and a link to the survey. This recruitment method enabled the inclusion of participants across multiple countries and with varied experiences, however, a response rate could not be calculated. Upon opening the survey link, participants were informed of the right to exit the survey at any time and gave voluntary consent by agreeing to complete the survey. To ensure participants could not be identified, names and email addresses were not collected with submissions.

### Instrument

The researcher designed an online survey utilizing Microsoft Forms to gather comprehensive data on

various aspects of participants' experiences related to CUULD. Potential questions were initially gathered from, and the survey was reviewed by members of a limb different occupational therapy practitioner group for clarity, relevance and content. Revisions were made based on their feedback prior to deployment of the survey.

The survey included four demographic questions regarding age, gender, geographic location, and specific location of the limb difference using rank ordinal scale.

Additionally, information about musculoskeletal conditions experienced and the impact on daily activities was collected using multiple response questions. Participants were asked to indicate and rate satisfaction with treatments they have previously received using a Likert-scale. Free response questions allowed for expansion on satisfaction with treatments received. In total, the survey consisted of 19 questions aimed at capturing the nuanced lived experience of the participants with CUULD.

### Procedures

Researchers employed an online survey as the primary data collection tool, disseminated through limb different specific groups on social media platforms as well as through individual social media pages. The survey commenced with participants reviewing the inclusion criteria and providing informed consent before proceeding to complete this comprehensive 19 question survey electronically. One reminder posting was made to the initial social media groups approximately six weeks after the original request.

### Data analysis

Following the completion of data collection, the survey data was downloaded onto an Excel version 2504 file where analysis of the results took place. Frequency analysis was completed, and measures of central tendency were calculated when applicable.

## Results

### Demographics

A total of 95 individuals actively participated in the survey. Demographic information is presented in [table 1](#). The demographic profile captured key characteristics, notably location of the limb difference, gender, age of respondent, and location of residency. 47 (52%) of respondents were under the age of 35 while the gender representation revealed a predominate female cohort, constituting 89.5% female ( $n = 85$ ) of the respondents, with male respondents accounting for 10.5% ( $n = 10$ ) of the sample. Most respondents indicated residing in the United States ( $n = 79$ ).

### Descriptive analysis

When asked, "Do you or have you experienced any of the following issues: pain, fatigue, carpal tunnel

**Table 1:** Characteristics of the study population.

Characteristic	Number (%) <sup>a</sup>
Location of Limb difference	
Partial finger/digit	8 (8)
Partial hand	38 (40)
Full hand	10 (11)
Between hand and elbow	24 (25)
At the elbow	6 (6)
Between elbow and shoulder	3 (3)
Full arm	5 (5)
Other	1 (1)
Gender	
Female	85 (89)
Male	10 (11)
Other options provided	0 (0)
Age of Respondent	
18 - 25	10 (11)
26 - 35	36 (41)
36 - 45	14 (15)
46 - 55	18 (19)
56 - 65	11 (12)
66 - 75	4 (4)
76 - 85	2 (2)
86+	0 (0)
Location of Residency	
United States	79 (83)
Europe	8 (8)
Asia	0 (0)
Africa	1 (1)
Australia/New Zealand	4 (4)
Canada	2 (2)
South/Central America	1 (1)
Age of Onset of Musculoskeletal Concern	
Under age 10	11 (12)
10 - 15	9 (9)
16 - 20	19 (20)
21 - 25	18 (19)
26 - 30	10 (11)
31 - 35	11 (9)
36 - 40	4 (4)
41 - 45	4 (4)
46 - 50	0 (0)
51 - 55	3 (3)
56 - 60	1 (1)
Over 60	0 (0)
No Concerns Reported <sup>b</sup>	6 (6)

**Note:** <sup>a</sup>n = 95; <sup>b</sup>Participants who did not report musculoskeletal concerns did not report an age of onset

syndrome, shoulder issues, swelling of an upper limb, or weakness in an upper limb?" 78 (82%) respondents (N = 95) answered yes.

Respondents were asked to indicate the age when their symptoms first started. Results included in [table 1](#) indicate that 60% (n = 57) of respondents reported experiencing symptoms such as pain, shoulder issues, weakness, etc. by the time they were 25 years old with 41% of those respondents (n = 39) experiencing symptoms by the age of 20.

Participants were asked to indicate location of symptoms on both the limb different side and the unaffected side. As shown in [table 2](#), respondents indicated pain, shoulder issues, carpal tunnel syndrome, tingling/numbness and weakness/fatigue in the limb different and/or the unaffected sides. Neck pain for both sides was reported with highest frequency (n = 48, 48) followed by general pain (n = 41) for the limb different side and tingling numbness (n = 40), shoulder issues (n = 39) and back pain (n = 39) for the unaffected side. Of note, respondents reported greater numbers for the unaffected side in all areas of concern except for pain.

The survey asked questions regarding the types of interventions they have received or currently use for physical issues related to their limb difference as well as the overall satisfaction of the interventions. [Table 3](#) demonstrates the percentage of respondents receiving the intervention and their satisfaction and dissatisfaction ratings of the interventions. As can be seen from the table, anti-inflammatory medications are the most frequently utilized interventions followed by physical therapy and massage.

Comparatively, however, surgical intervention, massage and chiropractic care were rated as most satisfactory. Of note, while utilized by 44 - 57% of

**Table 2:** Type of musculoskeletal concern by physical location.

	Unaffected Side (%)	Limb Different Side (%)
General pain	35 (37)	41 (43)
Neck pain	48 (51)	48 (51)
Back pain	39 (41)	37 (39)
Shoulder issues	39 (41)	28 (29)
Carpal tunnel	31 (33)	5 (5)
Tingling/numbness	40 (42)	22 (23)
Weakness/fatigue	36 (38)	34 (36)
No Concern Noted	17 (18)	14 (15)

**Note:** N = 95

**Table 3:** Receipt and satisfaction of interventions.

	Recipients Receiving <sup>a</sup> (%)	Recipients Satisfied <sup>b</sup> (%) <sup>c</sup>	Recipients Dissatisfied <sup>b</sup> (%) <sup>c</sup>
Anti-inflammatory Medications	54 (57)	14 (26)	7 (13)
Pain Medications	42 (44)	8 (19)	8 (19)
Occupational Therapy	39 (41)	17 (44)	7 (18)
Physical Therapy	52 (55)	21 (40)	7 (13)
Chiropractic Care	31 (33)	20 (65)	4 (13)
Massage	46 (48)	30 (65)	2 (4)
Surgery	33 (35)	23 (70)	2 (6)
Prosthetics	23 (24)	13 (57)	4 (17)

<sup>a</sup>n = 95; <sup>b</sup>Responses of satisfied and very satisfied are combined under satisfied and responses of dissatisfied and very dissatisfied are combined under dissatisfied; <sup>c</sup>Percentages of satisfaction and dissatisfaction derived from number receiving



**Table 4:** Activities impacted by physical issues experienced.

Activity	Number (%)
Weightlifting/strength training	49 (52)
Yard work/general maintenance	36 (38)
Exercise (walking, running, biking, etc)	33 (35)
Housekeeping	32 (34)
Exercise classes	27 (28)
Cooking/meal prep	24 (25)
Employment	20 (21)
Socialization with others	19 (20)
Daily Self Care	17 (18)
Driving	15 (16)
None Indicated	20 (21)

n = 95. Participants who did not respond to this question counted as none indicated.

respondents, pain and anti-inflammatory medications had the lowest overall satisfaction rates of 19 - 26%.

Participants were also asked about engagement in daily activities. When asked if they currently limit their engagement in activities due to the physical symptoms they experience, 49% (n = 47) answered 'yes'. Additionally, 38% (n = 36) indicated they have outright stopped participating in activities they once did due to physical symptoms. Table 4 details the activities impacted by the musculoskeletal issues experienced by the respondents. Of greatest impact is engagement in weightlifting and/or strength training followed by completion of yard work or general household maintenance activities.

## Discussion

This study aimed to determine the musculoskeletal concerns of individuals with CUULD and the impact on engagement in daily activities in depth. Additionally, the study sought to determine the types and satisfaction rates of interventions received by individuals with CUULD. The results of this study indicating 83% of respondents experience musculoskeletal concerns and 37 - 51% experience pain align with previous reports of pain in 63% of individuals with congenital limb differences [4]. Limited studies have explored the age of onset of overuse or pain in the limb different community however, onset of chronic pain in the general population has been found to be 14% - 23% of those aged 25 or younger [25,26]. The results of this survey indicate the age of onset before age 25 in 60% of respondents suggest pain and associated musculoskeletal issues present earlier for individuals with congenital limb difference. Given the impact of pain and musculoskeletal issues on function, prevention and treatment of these issues should be a priority for rehabilitative specialists working with the limb different population.

This study revealed that individuals with congenital limb difference limit their engagement in daily activities due to the musculoskeletal concerns they experience. This has also been reported in the literature indicating

impacts on employment and overall quality of life [15,17]. Additional research is warranted to determine the full impact of limb difference on engagement in daily activities as well as the influence of musculoskeletal concerns on engagement. Effective treatments aimed at engaging in preferred activities while reducing pain are needed.

The interventions with the greatest satisfaction reported were surgical interventions, massage and chiropractic care. While surgical intervention resulted in high satisfaction rates, only 35% of respondents reported having received surgery for issues related to their limb difference suggesting this is not a frequently prescribed or accepted intervention. These results are not surprising when considering the research indicating inconsistent surgical protocols and techniques for limb differences [20]. Responses to an open-ended question allowing for expansion on intervention satisfaction indicated that massage, chiropractic care providing soft tissue massage and targeted stretching are preferred. These results are consistent with research showing the benefits of deep tissue massage for those with myofascial pain syndrome which has features like those experienced by individuals with limb differences [27].

The variety of interventions reported by participants speaks to the need for multidisciplinary awareness of the issues faced by individuals with CUULD. The musculoskeletal impact of ULD is documented in literature and within this study with age of onset earlier than the general population. Limited research exists however on effective treatments available to these individuals as much of the literature to date primarily focuses on the care of children, surgical interventions and prosthetics.

## Limitations

Limitations for this study include the use of data collection through purposive and snowball sampling. Utilization of existing social media groups for individuals with limb difference could have resulted in limitations to the sample of participants as those not on social media were not made aware of the study. Additionally, most responses were received from females which could have impacted the experience of musculoskeletal issues and/or utilization of interventions given potential gender-based differences. The length of the survey could have resulted in response fatigue limiting the results of the later questions related to types of interventions and satisfaction levels as well as activities impacted by the limb difference.

## Recommendations for future research

This study underscores the necessity for expansion of multidisciplinary treatment and intervention approaches for individuals with CUULD. Additional research regarding specifics of interventions including type, frequency, duration and content are needed.

Multidisciplinary understanding of the impact of congenital limb difference on physical health and overall function will support further development of effective interventions and provide valuable support to individuals with CUULD.

## Conclusion

This study provides insight into the musculoskeletal concerns experienced by individuals with CUULD and underscores the need for multidisciplinary interventions. Through a comprehensive analysis of survey data from 95 participants, this study adds to the limited existing literature insights into the range of symptoms, activity limitations and treatment modalities received for this population. The findings reveal a variety of treatment options with variable satisfaction levels highlighting the need for further exploration of effective and accessible services to alleviate musculoskeletal concerns and promote participation in daily activities.

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## Author note

The author reports there are no competing interests to declare.

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