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Original Research

Activities and participation in young adults with Osteogenesis Imperfecta

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Abstract. The objective of this cross-sectional study was to compare the activities and participation in the domains of mobility, self-care, domestic life and social functioning in young adults according to osteogenesis imperfecta (OI) type. Fifty-four former OI patients were invited to participate and were sent a structured questionnaire. Twenty-four patients (mean age: 25.0 years, SD: 2.6 years) with OI types I ($n = 7$), III ($n = 7$), IV ($n = 8$) and V ($n = 2$) completed the questionnaire. Participants with OI type I reported full independence, and only few respondents with OI types IV and V reported some limitations in mobility and domestic life activities. Young adults with OI type III had significantly lower activity scores in aspects of mobility and domestic life and lower levels of participation in employment, sporting activities and transportation. Participation in leisure and social interactions were not different across OI types. Young adults with more severe types of osteogenesis imperfecta have greater activity limitations and participation restrictions. Our findings indicate the importance of promoting and facilitating involvement in meaningful activities and roles in young adults with moderate to severe forms of OI.

Keywords: Osteogenesis Imperfecta, activities and participation, outcomes

1. Introduction

Osteogenesis imperfecta (OI) is a heritable connective tissue disorder with an incidence of approximately one in 10000 births [7]. It affects males and females equally and occurs in all racial and ethnic groups. The principal clinical feature of OI is bone fragility, leading to frequent fractures [19]. Other clinical features may include blue sclera, dentinogenesis imperfecta (discoloration and brittleness of teeth), skin and ligamentous hyperlaxity, hearing impairments, presence of Wormian bones on skull radiographs, decreased height and skeletal deformities. In the majority of cases, OI is caused by mutations affecting collagen type I, the most prevalent protein in bone, skin and other connective tissues [19]. Cognitive impairment is not associated with this disorder [18].

OI is a very heterogeneous disorder. The severity of the skeletal involvement ranges from extreme bone weakness and death shortly after birth to absence of fractures. In an attempt to group patients with similar clinical features and disease severity together, David Sillence in 1979 devised a phenotypic classification that originally distinguished four types [21]. Subsequent work at our institution has led to the identification of three more clearly distinguishable types of OI [19]. Consequently, seven clinical types of OI are

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Table 1
Expanded Sillence classification of OI

Type	Clinical severity	Typical features	Typically associated mutations	Relative incidence*
I	Mild non-deforming OI	Normal height or mild short stature; blue sclera; no DI	Premature stop codon in COL1A1	47%
II	Perinatal lethal	Multiple rib and long-bone fractures at birth; marked deformities; broad long bones; low density of skull bones on x-rays; dark sclera	Glycine substitutions in COL1A1 or COL1A2	–
III	Severely deforming	Very short; triangular face; severe scoliosis; grayish sclera; DI	Glycine substitutions in COL1A1 or COL1A2	18%
IV	Moderately deforming	Moderately short; mild to moderate scoliosis; grayish or white sclera; DI	Glycine substitutions in COL1A1 or COL1A2	27%
V	Moderately deforming	Mild to moderate short stature; dislocation of radial head; mineralized interosseous membrane; hyperplastic callus; white sclera; no DI	unknown	4%
VI	Moderately to severely deforming	Moderately short; scoliosis; accumulation of osteoid in bone tissue, fish scale pattern of bone lamellation; white sclera; no DI	unknown	3%
VII	Moderately deforming to perinatal lethal	Severity ranging from death in first days of life to mild short stature. Short humeri and femora; white sclera; no DI	CRTAP	1%

Note: The ‘typically associated mutations’ may or may not be detectable in a given patient. DI: dentinogenesis imperfecta;
From: Rauch F, Glorieux FH. Osteogenesis imperfecta. Lancet 2004;363:1377-85.
*Patient population seen at the Shriners Hospital for Children in Montreal (total $n = 550$); patients with OI type II are usually not seen in rehabilitation/orthopedic environments, as they die shortly after birth.

distinguished at present (Table 1) [19]. OI type I is the most common type, with an incidence of almost 50%, and is associated with an increased risk of fracture but does not lead to deformities of long bones. OI type II is the most severe form of OI, resulting in death in the perinatal period caused by severe fractures and deformity of the thoracic cage. In children surviving the neonatal period, OI type III is the most severe form of the disease and occurs in about 20% of all patients with OI. It is characterized by severe progressive skeletal deformities with a high fracture incidence even through adulthood. OI type IV patients may exhibit mild to moderate bone deformities, variable short stature, dentinogenesis imperfecta and greyish sclera. The distinguishing features of OI type V are the occurrence of hyperplastic callus and a calcified interosseous membrane that limits forearm movement and can lead to radial head dislocation. OI types I through V are autosomal dominant disorders. OI type VI and VII are rarer forms of OI that are inherited in an autosomal recessive fashion.

Treatment of OI consists of rehabilitation including physiotherapy and occupational therapy to promote gross motor development and maximize functional independence, and orthopaedic surgery to stabilise bones and correct deformities. Bisphosphonate therapy is often used in patients with moderate to severe OI in an effort to increase bone mineral density, decrease fracture rates and alleviate pain [7,8]. The short term effects of these treatment modalities have been extensively studied, but most of these studies were limited to the analysis of body functions in pediatric OI populations.

In addition to measures of body function, the health and functioning of individuals can be described by additional domains as described by the International Classification of Functioning, Disability and Health (ICF). The World Health Organization (WHO) developed the ICF in 2001 in order to provide us with an international taxonomy for classifying and measuring function, disability, and health, using standard concepts and terminology across the lifespan [14,25]. The ICF views an individual’s health condition as being influenced by three domains: first, body functions and structures at the organ level; second, activities at the person level and, third, participation at the person-in-society level. Disability is viewed as the result of the complex relationship between an individual’s health condition and the factors specific to the person’s life that act as facilitators or barriers (Fig. 1). Activity is defined by the ICF as the execution of a task or action by an individual. Participation alludes to the involvement of an individual in a life situation and includes engaging in community life, recreation and leisure, and in religion and spirituality. Difficulties at the activity level are referred to as activity limitations, and difficulties an individual may experience in his/her involvement in life situations are referred to as participation restrictions [25].

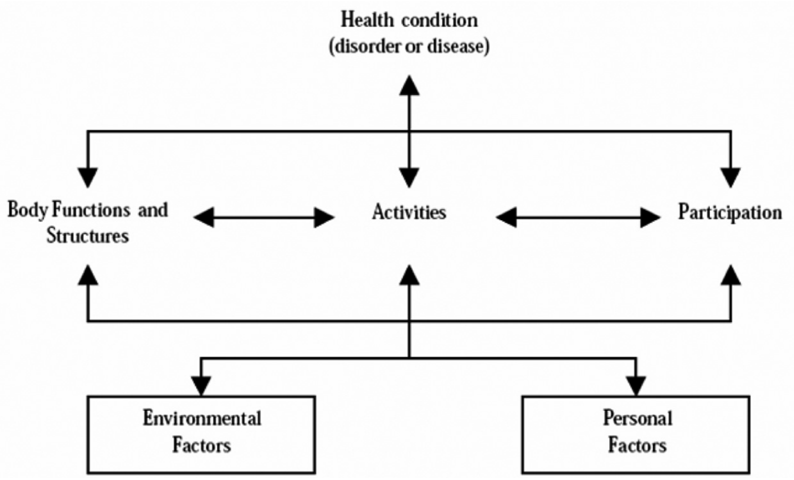

Fig. 1. The International Classification of Functioning, Disability and Health (ICF) WHO, 2001 (World Health Organization 2001).

Several studies [5,6,22,23] have examined physical function in children with OI, and an earlier study [20] has described their psychological functioning. These studies have focused on the body functions and body structures domains of the ICF, with limited to no attention to the activity and participation domains. In the adult population, two studies report on the functional outcome of adults with OI. Both studies [16,24], the first by Moorefield and Miller and the second by Widmann and colleagues, report high levels of educational achievement, as well as involvement in occupational activities, despite some participants having significant physical impairments. However, a limited range of activities and life roles were included in both studies, and although the latter study categorized participants into two OI categories, the results were not analyzed according to OI type. Currently, very little is known about the activities and participation in adults with OI according to OI type. Such information is required in order to evaluate the specific needs of young adults with varying functional skills and limitations. The objective of this study was to compare the activities and participation in the domains of mobility, self-care, domestic life and social functioning in young adults according to OI type.

2. Methods

The study population was comprised of individuals with a diagnosis of OI who were over 21 years of age, were no longer being followed at a pediatric facility and had previously received medical care at our center for a minimum of 5 years. This is a pediatric orthopedic hospital that provides multidisciplinary care for children and adolescents with OI, including orthopedic surgery, rehabilitation, and bisphosphonate therapy, depending on their individual needs [26]. Approval from the Director of Professional Services at the Shriners Hospital was obtained to conduct the chart review, in accordance with the guidelines provided from our Institutional Review Board. The ethics consultant at our institution considered this study to be within the realm of performance improvement. Therefore, IRB approval for contacting former patients by mail was not required and consent to participate was inferred by the return of completed questionnaires.

A review of the medical charts at the Shriners Hospital revealed 54 former OI patients who were 21 years or older at the time of the review. These individuals were invited to participate by mail and were sent a structured questionnaire. Twenty-four patients sent back the questionnaire and thus were included in this study. Demographic and diagnostic characteristics were similar between respondents and non-respondents (Table 2). The study population included 16 men and 8 women aged 21 to 33 years with OI types I, III, IV and V. There were no participants with types VI or VII.

The questionnaire used in this study is shown in Appendix I. It included questions on mobility, self-care and domestic life that are part of two different tools, the Functional Independence Measure (FIM) and the Instrumental Activities Measure (IAM). The FIM assesses physical and cognitive areas of dysfunction in activities which commonly occur in individuals with progressive, reversible or fixed neurologic, musculoskele-

Table 2
Demographic and diagnostic characteristics of respondents and non-respondents

Variable	Respondents ($n = 24$)	Non-respondents ($n = 25$)
Age, mean (SD)	25.0 (2.6)	24.4 (2.5)
Gender, n (%)		
Male	16 (67)	15 (60.0)
Female	8 (33)	10 (40.0)
OI type, n (%)		
Type I	7 (29)	5 (20)
Type III	7 (29)	5 (20)
Type IV	8 (33)	10 (40)
Type V	2 (8)	5 (20)

tal and other disorders [11]. The IAM was developed as a supplement to the FIM to provide information on patients' independence in instrumental activities of daily living and has been used in adults with spina bifida and cerebral palsy [2,10]. It is suggested that combining the FIM and IAM is useful for assessing persons with disabilities living in the community [10]. Interrater reliability of the FIM was found to be high; in the clinical setting an ICC of 0.96 for the total FIM was found, and item score agreement ranged from a Kappa of 0.53 (memory) to 0.66 (stair climbing) [12]. The reliability of the FIM was also examined using meta-analysis using 11 published studies, and found acceptable reliability across a wide variety of settings, raters and patients [17] including administering the FIM as a self-report questionnaire [13]. The FIM was also found to be more valid than the Barthel Index and equally reliable in the assessment of disability [15]. Interrater agreement of the IAM was good, with Kappa values exceeding 0.60 [4]. For practical reasons, 16 items from the FIM and IAM were included in the mailed questionnaire. These items addressed self care, domestic, and mobility related activities, which relate to the activity domain of the ICF. These 16 items were scored on a four-point ordinal scale: maximum assistance (1), moderate assistance (2), minimal assistance (3), and independent (4). In addition, the questionnaire also included 17 questions about participation in terms of education, employment, social interactions and leisure. These questions were developed by the members of the transition team of our hospital to investigate the issues and concerns that families and adolescents expressed during the interviews for the transition to adult health care. These items addressed education, work, and social roles which relate to the participation domain of the ICF. As well items on leisure activities were included and these relate to the activity domain of the ICF. Scoring for these questions ranged from dichotomous to 5-response options depending on the information being collected with the exception of question 17 which was open ended with multiple response options.

Descriptive statistics were calculated to describe the outcomes of young adults with OI and to examine differences in activity levels between OI types. Patients with OI types IV and V were treated as one group, because these OI types are similar in disease severity [9, 27] and the OI type V group was too small ($n = 2$) for comparisons on its own.

To determine whether participation levels varied between OI types, OI types were further classified into two groups, type I ($n = 7$), and types III, IV and V together ($n = 17$), which correspond to a mild form and a more severe form. Fisher's exact test (two-tailed) was conducted, with a 5% significance level.

3. Results

3.1. Activity scores (*Table 3*)

Young adults with OI type I all obtained the highest independence scores in all activity domains, whereas young adults with OI type III reported needing some assistance in all activities except for feeding. In comparison to the other OI types, young adults with type III seem to exhibit lower scores for transfers to car, simple meal preparation, cooking, cleaning and laundry. An intermediate picture emerged for those with OI types IV and V, who were independent in self-care activities but reported some difficulty in car transfers and outdoor mobility, as well as in the domestic life domain. The lack of variability in independence scores among those with type I precluded statistical testing, even after dichotomizing the outcome scores.

3.2. Participation (*Table 4*)

Significant differences between OI types were found with regard to participation. Compared to OI type III,

Table 3
Activity scores of young adults according to osteogenesis imperfecta (OI) type

Item, mean (SD)	OI type		
	Type I ($n = 7$)	Type III ($n = 7$)	Types IV + V ($n = 10$)
Self-care			
Feeding	4.0 (0.0)	4.0 (0.0)	4.0 (0.0)
Grooming	4.0 (0.0)	3.6 (0.7)	4.0 (0.0)
Bathing	4.0 (0.0)	3.4 (1.1)	4.0 (0.0)
Dressing upper body	4.0 (0.0)	3.4 (1.1)	4.0 (0.0)
Dressing lower body	4.0 (0.0)	3.3 (1.2)	4.0 (0.0)
Toileting	4.0 (0.0)	3.4 (0.9)	4.0 (0.0)
Transfers/outdoor mobility			
Transfer to toilet	4.0 (0.0)	3.3 (1.2)	4.0 (0.0)
Transfer to bath	4.0 (0.0)	3.3 (1.2)	4.0 (0.0)
Transfer to chair	4.0 (0.0)	3.6 (0.7)	4.0 (0.0)
Transfer to car	4.0 (0.0)	2.7 (0.9)	3.9 (0.3)
Mobility outdoors	4.0 (0.0)	3.7 (0.8)	3.7 (0.6)
Domestic life			
Simple meal preparation	4.0 (0.0)	2.9 (0.6)	3.9 (0.3)
Cooking	4.0 (0.0)	2.6 (0.9)	3.6 (0.9)
Shopping	4.0 (0.0)	3.3 (1.0)	3.4 (1.0)
Cleaning	4.0 (0.0)	2.0 (0.5)	3.5 (0.9)
Laundry	4.0 (0.0)	2.0 (1.1)	3.5 (1.0)

4 = Independent; 3 = Minimal assistance; 2 = Moderate assistance; 1 = Maximum assistance.

Table 4
Participation of young adults according to osteogenesis imperfecta (OI) type

Participation variable, n (%)	OI type		p-value
	I ($n = 7$)	III/IV/V ($n = 17$)	
Education			
• Attending school	1	8	0.191
• Not attending school	6	9	
Employment			
• Working	7	8	0.022
• Not working	0	9	
Volunteer work			
• 1–10 hours/week	1	3	1.00
• Not volunteering	6	14	
Living arrangements			
• Living alone	2	7	0.699
• Living with other	5	10	
Sports activities (such as walking, hiking, swimming, basketball, bike riding)			
• Weekly/Monthly	6	5	0.023
• Never	1	12	
Social activities outside the home (church groups, shopping or movies with friends)			
• Regularly/Occasionally	5	17	0.076
• Never	2	0	
Leisure activities (reading, crafts, TV, music, computer)			
• Regularly/Occasionally	7	17	NA
• Never	0	0	
Main transportation in the community			
• Drives car	6	7	0.078
• Uses other transportation mode (passenger/public transport/adapted transport)	1	10	

IV and V, more young adults with OI type I were working and participated in sporting activities. Nine of the participants reported living on their own; one of them with supervision. Fifteen of the participants reported various living arrangements including two living with their partner and one living with their child. All participants reported that they engaged in social and leisure activities at least occasionally.

Open-ended questions enquired the areas where help is needed to increase patients' independence; the top three areas identified were housing/vehicle adaptations, driver's education and help getting a job.

4. Discussion

The present study shows that engagement in certain activities and life situations in young adults with OI vary according to OI type. All participants with OI type I reported full independence, and only a few respondents with OI types IV and V reported needing minimal assistance in mobility and domestic life activities. In contrast, young adults with OI type III had lower activity scores in some aspects of mobility and domestic life. The severe short stature and upper extremity deformity associated with OI Type III may contribute to the restrictions experienced in mobility, transfers, cooking and laundry. Participants with OI types III, IV and V also had lower levels of employment and participation in sporting activities. A childhood of bone fragility may explain these participants' reluctance to become involved in sports. Interestingly, living arrangements varied among the participants and were not related to type of OI. As well, participation in leisure and social interactions were not different across OI types. The lack of differences in these areas across OI types may be explained by the age appropriate social skills and normal cognition associated with OI. These personal factors then act as facilitators.

Our findings are comparable to a study [24] on 30 adults with OI which reported a high percentage of ability in the activities of daily living (ADL) subset of the FIM, with only seven of the 23 patients exhibiting a deficit in the ADL subset. The authors conclude that the study population was very functional despite having musculoskeletal limitations. However, function was assessed unidimensionally, using only a daily living subset, and do not reflect function in more complex activities and performance in life roles. It is also expected that those patients reporting a deficit have a more severe type of OI; however this was not reported. An early study by Moorefield and Miller [16] on 31 adults with OI (mean age = 29 years) following surgical treatment found that 5 were ambulating without technical aids, 18 were walking with braces and/or crutches, and 8 remained non-ambulatory post-operatively. In terms of education and employment, 21 had graduated from or were attending high school, and 20 were employed. Two-thirds enjoyed hobbies and recreational activities involving social contacts. Eighteen patients drove, including five which needed hand controls. These results indicate that adults with OI were for the most part productive despite physical limitations. However, a limited range of activities and life roles were assessed which limited the identification of their needs in these areas.

Our results appear to indicate that young adults with more severe types of OI have greater limitations in more physically complex self-care activities, such as grooming, bathing, dressing, and toileting. Indeed, young adults with type III OI reported needing moderate assistance on average for outdoor mobility and transfers, and all activities of domestic life. Nevertheless, many of the respondents are living similarly as expected for young adults even though they experience these aforementioned difficulties.

OI is a rare disease with several subtypes. In contrast to the earlier studies our study attempted to compare levels of activity and participation according to the latest classification. The small sample size in this study precluded further statistical analysis to explore which factors are associated with independence in the activity and participation domains. In addition, the FIM and IAM was not discriminative enough within OI types (maximum scores were obtained in all items by participants with type I OI) providing no variation and thus precluding statistical analysis. The multiple tests in this study were not independent and correlated (employment and volunteer work), therefore using a Bonferonni correction was inappropriate as it is highly conservative and may miss real differences [3].

Studies with a larger sample size permitting comparison between different OI types and using validated measures of activities and participation capable of discriminating among OI types are needed. Furthermore environmental factors such as access to programs for adapting residences and vehicles may influence involvement in life roles and therefore should also be explored in future research. Qualitative type research has rarely been used in this population, but is indicated to explore the individual experiences of young adults with OI and to understand which areas require intervention to maximize independence and well-being in adulthood. This study demonstrates the importance of understanding the level of performance of individuals with OI in activity and participation domains. Our findings indicate the need to promote and facilitate involvement in meaningful activities and roles in young adults with moderate to severe forms of OI.

5. Clinical messages

- Young adults with more severe types of OI have greater limitations in self-care, mobility and domestic activities, are less employed, and participate less in sports than young adults with mild involvement.

- No differences were noted in participation levels in leisure and social activities among different OI types. Many respondents are living similarly as expected for young adults, even though they experience difficulties with self-care, mobility, and domestic activities.
- Our findings indicate the importance of promoting and facilitating involvement in meaningful activities and roles in young adults with moderate to severe forms of OI.

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Appendix 1- Sample questionnaire

To be completed by former Shriners Hospital patient

1. Are you in school now?
   ☐ No, please describe level of education completed ________________
   ☐ Yes, please describe____
2. Have you completed any additional training for specific skills outside of school?
   ☐ No
   ☐ Yes, please describe____
3. a) Are you working for an income now?
   ☐ No
   ☐ 1 to 10 hours per week
   ☐ 11 to 25 hours per week
   ☐ 26 to 40 hours per week
   Please describe what you do at work____
3. b) If no, Do you want to work? Yes ☐ No ☐
   Are you looking for work? Yes ☐ No ☐
3. c) What would help you be able to work?
   ☐ Transportation
   ☐ Special adaptations
   ☐ More education or training
   ☐ Getting over my fear of working
   ☐ My parents learning to let go
4. Do you do volunteer work in your community?
   ☐ No
   ☐ 1 to 10 hours per week
   ☐ 11 to 25 hours per week
   ☐ 26 to 40 hours per week
   Please describe:____
5. What are your current living arrangements?
   ☐ Family home
   ☐ Supervised apartment
   ☐ Living with siblings or friends in community
   ☐ Living with partner
   ☐ Other, please describe____
6. Do you participate in sports activities? (Such as walking, hiking, swimming, basketball, bike riding)
   ☐ Never
   ☐ 1 or 2 times per month
   ☐ 1 or 2 times per week
   Please describe:____
7. Do you participate in social activities outside your home? (Church groups, shopping or movies with friends)
   ☐ No
   ☐ Occasionally
   ☐ Regularly
   Please describe:____
8. Do you participate in leisure activities? (Reading, crafts, TV, music, computer)
   ☐ Never
   ☐ Occasionally
   ☐ Regularly
   Please describe____

9. How do you travel in the community? (Check as many as apply)
   ☐ Drive your own car
   ☐ Passenger in family car
   ☐ Use adapted transport services on your own
   ☐ Use public transportation, If yes, please indicate how
       ☐ Independently
       ☐ With minor assistance
       ☐ With moderate assistance
       ☐ With maximum assistance
10. In general how would you rate your health?
   ☐ Excellent
   ☐ Very good
   ☐ Good
   ☐ Fair
   ☐ Poor
11. Do you receive medical follow-up in your area? (Family doctor, clinic, hospital)
   ☐ No, please comment________________________________
   ☐ Yes, please describe________________________________
       ☐ Family doctor
       ☐ Orthopedics
       ☐ Other
       When was your last visit:________________________________
12. Does this doctor know how to manage your condition/special needs?
   ☐ No
   ☐ Yes
   Please describe:____
13. Are you able to get what you need to manage your health condition? (medication, mobility equipment, etc.)
   ☐ Yes
   ☐ No, please comment________________________________
14. Do you talk with your doctor /other health care providers yourself?
   ☐ All the time
   ☐ Sometimes
   ☐ Never
15. Do you make your own decisions about your treatment plan?
   ☐ All the time
   ☐ Sometimes
   ☐ Never
16. Do you:
   ☐ Use a wheelchair at all times
   ☐ Walk with aids at all times
   ☐ Use a combination of wheelchair and walking
   ☐ Walk without aids at all times
17. What do you need to help you increase your independence?
   ☐ Transportation
   ☐ Drivers education
   ☐ Education
   ☐ Personal care attendant
   ☐ Housing or vehicle modifications
   ☐ Help to get a job
   ☐ Help managing your health condition
   ☐ Community recreation

☐ Counselling/support groups
☐ More experience making decisions
☐ Nothing
☐ Other______________________________________

18. Please rate your level of independence in the following areas:

	Independent	**Minimal Assistance**	**Moderate Assistance**	**Maximum Assistance**
Feeding (using fork or spoon, cup)	☐	☐	☐	☐
Grooming (brushing hair, teeth, washing and drying face & hands)	☐	☐	☐	☐
Bathing (Washing and drying body/neck down)	☐	☐	☐	☐
Dressing Upper Body (T-Shirt, jacket buttons)	☐	☐	☐	☐
Dressing Lower Body (pants, socks, shoes, braces)	☐	☐	☐	☐
Toileting (manages clothing and wiping self)	☐	☐	☐	☐
Transfer to toilet	☐	☐	☐	☐
Transfer to bath	☐	☐	☐	☐
Transfer to chair	☐	☐	☐	☐
Transfer to car	☐	☐	☐	☐
Mobility Outdoors (walking or using wheelchair in residential area)	☐	☐	☐	☐
Simple Meal Preparation (making sandwiches, cup of tea/coffee)	☐	☐	☐	☐
Cooking (making a meal for 1-2 persons, including washing dishes)	☐	☐	☐	☐
Shopping (going to stores, choosing articles, paying, carrying articles home)	☐	☐	☐	☐
Cleaning (making bed, vacuuming, daily tidying)	☐	☐	☐	☐
Laundry (sorting, using washing machine, folding, etc)	☐	☐	☐	☐

19. Please rate how well the Shriners Hospital helped you to:

	Greatly	Moderately	Minimally
• Stay healthy and independent	☐	☐	☐
• Find adult health care	☐	☐	☐
• Learn to make your own decisions	☐	☐	☐

20. Can you share with us some of your future plans____

General Comments____

Thank you for your participation!