

Osteogenesis imperfecta: Level of independence and of social, recreational and sports participation among adolescents and youth

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ABSTRACT

Introduction. Osteogenesis imperfecta is a group of hereditary connective tissue disorders that cause bone fragility, with a wide clinical variability resulting in varying degrees of motor disability.

Objectives. To describe the level of independence and of social, recreational and sports participation among adolescents with osteogenesis imperfecta.

Population and methods. Descriptive, analytical and cross-sectional study conducted in patients with osteogenesis imperfecta older than 15 years old attending the Skeletal Dysplasia Office of Hospital "Prof. Dr. Juan P. Garrahan" (May 2013 through December 2014). Self-administered survey. Short stature was an outcome measure that indicated severity. **Results.** There were 18 patients; age: 19.17 (± 3.4 sDE); 83% had moderate-severe forms of OI; median height: -7.9 sDE; 50% used a wheelchair.

Average education years: 12.2; 56% participated in sporting activities; and 78% were involved in recreational and social activities. A high level of independence was observed.

We found a correlation between short stature and use of wheelchair ($r: -0.77$) and between short stature and participation in sporting activities ($r: 0.66$). No correlation was observed with years of education ($r: -0.15$), participation in social activities ($r: -0.22$) or recreational activities ($r: 0.35$).

Key words: *osteogenesis imperfecta, adolescent, social participation, recreational activities.*

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INTRODUCTION

Osteogenesis imperfecta (OI) is a group of hereditary connective tissue disorders that cause bone fragility.¹ According to its etiology, this

disease is directly or indirectly related to type I collagen,² and has a prevalence of 1 in every 12 000-15 000 newborn infants (NBIs).³

It has a wide clinical variability. The original classification made by Silience in 1979 into type I, II, III and IV was modified by Dijk and Silience in 2014 based on the clinical severity of OI into mild, moderate, severe and extremely severe.¹ The severe forms usually show multiple fractures observed before birth and lead to skeletal deformities and complications. Other common signs include variable degrees of short stature, dental alterations and hearing loss. OI is not associated with intellectual disability.⁴

There is no single therapeutic scheme for OI; management should be adapted to each individual and agreed among the multidisciplinary medical team, the patient and his/her family.^{3,5}

A study conducted in 2011 at the Shriners Hospital for Children Canada that caters for patients with OI conducted a survey to assess the level of independence and of social, recreational and sports participation among 24 adolescent and young adult patients with OI.⁴

We believe that having a broader picture of these aspects regarding our group of patients with OI may help us improve treatment strategies.

OBJECTIVE

To describe the level of independence and of social, recreational and sports participation among youth with OI.

POPULATION AND METHODS

This was a descriptive, analytical, cross-sectional study.

All patients with OI, older than 15 years old and attending the Skeletal Dysplasia Office of Hospital "Prof. Dr. Juan P. Garrahan" (May 2013 through December 2014) were assessed. The following clinical outcome measures were evaluated: age, sex, type of OI, zH. The latest classification published by van Dijk¹ was used. All patients were invited to complete a self-administered survey, which had been translated

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from the version prepared by the Shriners Hospital for Children Canada.⁴ The translation was done by one of the authors and then tested in three patients for comprehension, after which minimal grammar corrections were made. Before the survey was completed, patients gave their verbal consent to participate.

The survey included 20 questions. Questions 1-17 and 19 asked about education and work achievements, housing, transportation, relationship with the hospital and social, recreational and sports participation. Questions were close-ended, with single or multiple choice answers, depending on the question. Question 18 assessed 15 items regarding mobility, self-care and activities of daily living. These items were scored using a four-point scale (from requiring maximal assistance to being independent) and were selected from two different instruments: the Functional Independence Measure (FIM) and the Instrumental Activities Measure (IAM).⁶ The FIM assesses physical and cognitive impairment. The IAM was developed as a supplement to the FIM to obtain information on the level of independence. Question 20 was open-ended and asked about future plans. A copy of the questionnaire is attached in the *Annex*.

The measured height Z score (zH) was used as an outcome measure that indicated severity (a shorter stature meant a worse severity). The independent outcome measure used for correlations was zH. A correlation with the following selected outcome measures from the survey was sought: use of wheelchair, participation in social activities (involvement in religious groups, going to the mall or watching movies with friends, etc.), sporting activities (walking, swimming, playing basketball, etc.), recreational activities (reading, handicrafts, computer activities, etc.), and number of completed school years (completed years of mandatory education: as of 5 years old).

Statistical analysis

Data were summarized using central tendency and dispersion, as applicable. Spearman's correlation coefficient for nonparametric outcome measures was used to correlate zH to the different selected outcome measures.

RESULTS

General characteristics of the sample

All patients agreed to complete the questionnaire, which was self-administered by

all except one patient who required help due to his motor disability.

Eighteen patients with osteogenesis imperfecta older than 15 years old were included (11 boys); their mean age was 19.17 (± 3.4 sDE).

Of them, 83% had moderate-severe OI and 17%, mild OI. Median zH was -7.9 sDE (-14.68/+1.5 sDE).

Questionnaire analysis

To move around, 50% of patients used a wheelchair all the time, 17% combined a wheelchair with a walking aid, and 33% walked on their own.

In relation to education level, work, housing and transportation, the average number of completed school years was 12.2, and 78% were in school. Among patients older than 18 years old (11 patients), 36% were working, 69% would like to work, and 24% believed that they required adaptations. Also, 89% lived in their family house, and 78% used public transportation, but only 29% did so in an independent manner. Only one patient drove his own car. Fifty percent got driven the family car.

Participation in social, recreational and sporting activities is described in *Table 1*.

In relation to the perception of their own health status, their ability to manage OI and communication with the medical team, as well as their interaction with the health system outside our hospital, 55% of patients indicated that they had an excellent health status; 39%, that it was good; and only 1 patient, that it was fair. Sixty-six percent of patients received care close to their home, and 50% referred that their doctor had qualifications to manage OI. Also, 94% of patients felt capable of managing their condition, but 61% discussed their medical needs with their doctors only sometimes and 43% were always involved in making treatment decisions.

TABLE 1. Participation in sporting, social and recreational activities among adolescents with osteogenesis imperfecta (N: 18)

Participation	Regularly	Occasionally	Never
Sporting	39%	17%	44%
Social	50%	28%	22%
Recreational	67%	11%	22%

When asked to what extent our hospital had helped them feel healthy and independent, 95% of assessed patients indicated that it did to a great extent. In addition, 61% of patients indicated that the hospital helped them to make their own decisions to a great extent, but only 46% referred that it helped them to a great extent to find a hospital where they could receive adult care.

The level of independence in relation to mobility, self-care and activities of daily living is described in *Table 2*.

Participants were invited to share their plans for the future, and all but one wrote about them. The most common plans for the future included a wish to start or complete a university degree, work and have their own family.

Correlation between short stature and the different outcome measures

A short stature measured as Z score was used as a severity outcome measure. A negative correlation was observed between zH and wheelchair requirement ($r: 0.77$). The group with the higher level of participation in sporting activities was the one less affected by short stature ($r: 0.66$). No correlation was observed between zH and years of education ($r: 0.15$), participation in social activities ($r: -0.22$) or recreational activities ($r: 0.35$).

DISCUSSION

Weaknesses of our work include that the survey was not transculturally adapted and that

the sample size was not large; however, given that OI is a rare disease, we believe that the resulting data may be useful.

It was observed that, to varying degrees, 78% of patients participated in social and recreational activities and no correlation was found between zH and the frequency of such participation. Data published by Montpetit⁴ are similar to our results: no differences were observed in social and recreational involvement among the different types of OI. Compared to those activities, sports were practiced to a lower extent, consistent with Montpetit's reports, who observed that patients with severe forms of OI practiced fewer sporting activities.⁴

The levels of independence as described by mobility, self-care and activities of daily living are high. Difficulties are consistent with those observed among patients with type III OI in the referenced publication.⁴

Cole⁸ suggested that, although OI is associated with a normal intelligence, it may have an impact on academic performance given its major physical involvement, pain and motor disability. In our group of patients, no correlation was observed between the number of completed school years and severity, consistent with the high level of academic achievement observed in other populations with OI as reported by Montpetit⁴ and Widmann.⁹

The review conducted by Thompson¹⁰ among adults with skeletal dysplasia described varying degrees of social isolation and its influence on the

TABLE 2. Level of independence in relation to mobility, self-care and activities of daily living among patients with osteogenesis imperfecta (N: 18)

	Independent	Needs minimal assistance	Needs moderate assistance	Needs maximal assistance
Feeding	16	1	1	0
Personal hygiene	14	3	1	0
Bathing/showering	15	1	1	1
Dressing the upper body	18	0	0	0
Dressing the lower body	16	0	1	1
Transfer to the toilet	11	1	1	5
Transfer to the tub/shower	11	2	0	5
Transfer to the wheelchair	11	3	1	3
Transfer to the car	10	2	1	5
Mobility outside the house	14	1	2	1
Preparing simple meals	15	1	1	1
Cooking and doing the dishes	9	2	1	6
Shopping	14	1	1	2
Cleaning	9	3	1	5
Doing the laundry	10	1	2	5

level of education attained. Attention should be paid to this aspect given that, in our study, 22% of patients referred that they never participated in social activities.

Future studies should focus on establishing how the level of independence and participation may affect the quality of life of patients with OI.^{11,12} ■

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ANNEX

First and last name:
Date of birth:
Medical record no.:
Contact number:
Date of visit:

QUESTIONNAIRE

- 1) Are you in school this year?
No.
Yes.
School grade:
Type of education (standard, special, home-school):
Did you repeat or miss a school year? Please describe:
- 2) Have you completed courses outside school?
No.
Yes. Please describe:
- 3.a) Are you working for a salary?
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 you are not working, would you like to work?
Yes.
No.
Are you looking for a job?
Yes.
No.
- 3.c) What would help you have a job?
Transportation.
Special adaptations.
Better education or training.
Overcoming the fear to work.
My parents' approval.
- 4) Have you done community work (unpaid group work done for the benefit of the community)?
No.
1 to 10 hours per week.
11 to 25 hours per week.
26 to 40 hours per week.
Please describe:
- 5) What is your housing situation?
Living in my family home.
Living in a supervised apartment.
Living with friends.
Living with a couple.
Other. Please describe:

- 6) Do you participate in sporting activities (walking, camping, swimming, playing basketball, others)?
Never.
Occasionally (once or twice a month).
Regularly (once or more times a week).
Please describe:.....
- 7) Do you participate in social activities outside home (religious groups, going to the mall with friends, watching movies with friends, etc.)?
Never.
Occasionally (once or twice a month).
Regularly (once or more times a week).
Please describe:
- 8) Do you participate in recreational activities (reading, handicrafts, computer activities)?
Never.
Occasionally (once or twice a month).
Regularly (once or more times a week).
Please describe:
- 9) How do you travel around your community?
Driving your own car.
Getting driven in the family car.
Using a special transportation service.
Using public transportation.
If you use public transportation, indicate how:
On your own (you do not need assistance).
With minimal assistance (you need some help).
With moderate assistance (you need moderate help).
With maximal assistance (you need a lot of help).
- 10) In general, how would you describe your health status?
Excellent.
Very good.
Good.
Fair.
Poor.
- 11) Do you get medical care close to your home?
No. Please explain:.....
Yes. Please describe:
Primary care physician.
Traumatologist.
Other.
When did you last see your doctor?
- 12) Does your doctor have the qualifications to manage osteogenesis imperfecta?
Yes.
No.
Please describe:
- 13) Are you capable of taking care of yourself given your osteogenesis imperfecta?
Yes.
No. Please explain:

- 14) Do you discuss your needs with your doctors?
 All the time.
 Sometimes.
 Never.

- 15) Do you make your own decisions regarding your treatment plan?
 All the time.
 Sometimes.
 Never.

- 16) How do you move around?
 Using a wheelchair all the time.
 Using a wheelchair and walking.
 Walking with an aid all the time.
 Walking on your own all the time.

- 17) What would help you increase your independence?
 Transportation improvements.
 Learning to drive.
 Education.
 A personal care assistant.
 House or car modifications.
 Help to get a job.
 Help to manage your health condition.
 Community involvement.
 Support group.
 More experience to make decisions.
 Nothing in particular.
 Other:

18) Indicate your level of independence: (*Table*)

	Independent	Needs minimal assistance	Needs moderate assistance	Needs maximal assistance
1. Feeding (using cutlery and a glass).				
2. Personal hygiene (combing your hair, brushing your teeth, washing your hands and face).				
3. Bathing/showering (washing and drying your body).				
4. Dressing your upper body (putting on a tee shirt, buttoning a jacket).				
5. Dressing your lower body (putting on pants, socks, shoes, underwear).				
6. Transfer to the toilet (sitting down on the toilet).				
7. Transfer to the tub/shower (getting in the tub/shower).				
8. Transfer to the wheelchair (sitting down on your wheelchair).				
9. Transfer to the car (sitting down on the car seat).				
10. Mobility outside the house (walking or using a wheelchair outside your house).				

	Independent	Needs minimal assistance	Needs moderate assistance	Needs maximal assistance
11. Preparing a simple meal (making a sandwich, a cup of tea or coffee).				
12. Cooking (preparing food for 1 or 2 people, including doing the dishes).				
13. Shopping (going to the store, picking items, taking items home).				
14. Cleaning (making the beds, vacuuming, keeping the house clean).				
15. Doing the laundry (sorting out clothes, using the washing machine, folding the clothes).				

19) Please indicate how Hospital Garrahan has helped you with the following:

To a great extent To a moderate extent To a minimum extent

- Staying healthy and being independent.
- Finding a place where you can receive adult care.
- Learning to make your own decisions.

20) Would you share some of your future plans with us?.....
.....
.....
.....

Thanks for participating