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Osteogenesis imperfecta: towards an individualised interdisciplinary care strategy to improve physical activity and quality of life

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Summary

BACKGROUND: This report describes a new strategy for the care of patients with osteogenesis imperfecta, based on an interdisciplinary team working. Thereby, we aim at fulfilling three main goals: offering thorough coordinated management for all, and improving physical activity and quality of life of the patients.

AIM: With rare diseases such as osteogenesis imperfecta (OI), patients and their family often suffer from inadequate recognition of their disease, poor care coordination and incomplete information. A coordinated interdisciplinary approach is one possible solution for providing both comprehensive and cost-effective care, with benefits for patient satisfaction. Poor physical activity and impaired quality of life represent a considerable burden for these patients. To better address these issues, in 2012 we created an interdisciplinary team for the management of OI patients in our University Hospital Centre (CHUV, Lausanne University Hospital,). In this article we describe the implementation of this interdisciplinary care strategy for patients suffering from OI, and its impact on their physical activity and quality of life.

METHODS: All patients from the French part of Switzerland were invited to join us. We proposed two complementary evaluations: the initial interdisciplinary evaluation and a yearly follow-up during a special day – the "OI day". This day features specialised medical appointments adapted to each patient's needs, as well as lectures and/or workshops dedicated to patients' and families' education. Our first aim was to propose for each patient the same management, from diagnosis to the bone health evaluation and physical therapy advice. Our second aim was to evaluate the evolution of physical activity, quality of life (measured by EQ-5D, SF-36 and a dedicated questionnaire) and satisfaction of patients and their families. Here we report both the initial and the long-term results.

RESULTS: Since 2012, 50 patients from the French part of Switzerland received the personalised medical evaluation. All of the patients included in this study had the same initial evaluation and at least one participation in an OI Day. All patients had an adaptation of their bone acting drugs. Over a 7-year period, 62% of inactive patients started some physical activity, and 44% of patients who were not involved in any athletic activity started participating in sports. The mean EQ-5D increased from 0.73 to 0.75 (p = 0.59). The mean physical SF36 (musculoskeletal function) score was 59.09 ± 22.72 and improved to 65.79 ± 21.51 (p = 0.08), whereas it was 68.06 ± 20.05 for the mental SF36 without alteration during follow-up. The OI day was revealed to be useful, it contributed to improvement in continuity of care and helped families to better understand the OI patients' health.

CONCLUSIONS: Our interdisciplinary approach aimed at offering the same thorough management for all patients from the French part of Switzerland, and at improving both the physical activity and the satisfaction of the patients and their family. This report is a basis for future work focusing on the effect of bone fragility and the impact of OI on patients' social relations.

Keywords: *osteogenesis imperfecta*, *interdisciplinary*, *rare bone disease*, *physical function*, *quality of life*

Introduction

Osteogenesis imperfecta is a rare genetic connective tissue disorder with a wide phenotypic and molecular heterogeneity. The characteristic features and severity of OI vary greatly from person to person [1]. Skeletal fragility results in an increased risk of bone fracture in early life and progressive bone deformities, and extra-skeletal manifestations can lead to dental alterations (dentinogenesis imperfecta) and/or hearing impairment [2, 3]. Despite expanding research into its treatment, OI remains a burden that limits

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Data acquisition: All. Data interpretation: BAR, LB, BCX and AB. Drafting manuscript: BAR, CR and AB. Revising manuscript: all authors. Approving final version of manuscript: all authors. BAR affirms that the manuscript is an honest, accurate and transparent account of the study being reported.

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a person in daily activities and significantly impairs quality of life [4–6].

Over the last decade, numerous efforts have been made, with considerable emphasis, by national institutions, leading to the recognition of rare diseases by the European Union (EU). The "concept of rare disease" was acknowledged by the Swiss Federal Council in 2014, triggering its recognition and implementation in the 2020 Health Strategy Plan. This "concept" highlights intensive efforts to achieve global care for patients with a rare disease. Creating reference centres, enhancing care, adopting an interdisciplinary approach, educating patients and their loved ones, and increasing funding for research and treatment are all desired goals.

By adopting an interdisciplinary approach, we aimed at sharing information and educating patients and family members, as well as recent knowledge and therapeutic advances. We hypothesised that such a global "concept" would improve patients' management and outcomes, including musculoskeletal health, physical activity and quality of life [7–9]. The interdisciplinary approach for OI patients was created in 2012 (referred as to the "OI CHUV" group), with two primary goals: (1) to provide each and every patient with the same initial evaluation, the same access to genetic testing and the same follow up; (2) to assess the impact of this OI CHUV management on patients' physical activity and quality of life. Here we report our first results after 7 years of follow up, including a survey addressing patients' and families' satisfaction.

Material and methods

Study design and setting

Between October 2012 and October 2019, all patients attending an OI-dedicated consultation in a single Swiss centre (Bone and Joint Department, CHUV, Lausanne University Hospital, Switzerland) and seen by one member of the OI team could be included in this prospective study. Informed consent was obtained from each patient or caregiver. This study and its related prospective data collection were approved by the Ethics Committee of Vaud (Switzerland, CER Vaud 2018-01673).

Sample

Any patient, whether adult or child, referred to the CHUV (Lausanne University Hospital, Lausanne, Switzerland) for a suspected or confirmed OI diagnosis, was prospectively included in the OI CHUV group cohort. To be included in the present report, patients had to have attended at least one "OI day" (described below) since 2012.

Structure and development of the interdisciplinary group in the CHUV for all patients with OI (adults, children)

Before the recognition of OI as a rare disease by the Swiss Council, the OI days and OI CHUV team were created in 2012, in our tertiary hospital: the CHUV. At first, the interdisciplinary team was composed of two bone disorder specialists (an adult and a child specialist), one orthopaedic surgeon, one geneticist, and three physiotherapists (two for adults and one for children). Then, in 2013, a dentist joined the team, followed by an otorhinolaryngologist in 2016. Each team member had expertise in clinical OI management. However, by integrating a more global OI network, each specialist shared the same goal: more effective decision making and treatment. All health workers involved had a strong culture of team working, which allowed constructive discussion and optimised coordination of the different agendas. Such information was conveyed through local and national professional networks [4, 10] associations (The Swiss Association of Patients with OI) and the internet (https://www.info-maladies-rares.ch/).

Same Initial evaluation for all patients with suspected OI

All patients underwent a complete physical examination to confirm the clinical diagnosis of OI and to determine its phenotype according to the Sillence classification system [11, 12] (fig. 1).

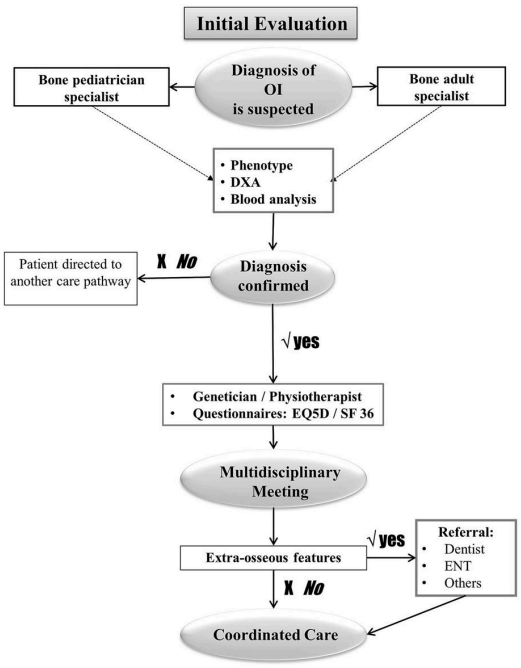
Assessment of the physical activity

Each patient was assessed for musculoskeletal and respiratory disorders (spirometry) by a physical therapist. A questionnaire was used to evaluate patient's physical practice (physical activity and sport), and each patient was counselled on physical and athletic activities and provided with recommendations for physical therapy.

Assessment of the quality of life

Quality of life refers to an interdisciplinary concept that varies across subject and time. It encompasses multiple notions pertaining to the physical, emotional and social domains [13]. Although discrepancies remain as to which dimensions to include or not in a quality of life evaluation, we chose to use the French version of the EQ-5D [14, 15], a widely used and validated tool for estimating health status preferences (utilities). The EQ-5D questionnaire allows

Figure 1: Initial evaluation pathway for OI patient in the CHUV OI group.; CHUV = Centre Hospitalier Universitaire Vaudois; DXA = dual X-ray absorptiometry; OI = osteogenesis imperfecta



assessment of five dimensions (mobility, self-care, usual activities, pain/discomfort and anxiety/depression) with three response options for each item: 1 = no problems; 2 = some problems; or 3 = severe problems, resulting in 243 possible health states. The instrument is complemented by a visual analogue scale that allows individuals to rate their overall health status on a scale from 0 to 100. The visual analogue scale provides a purely patient-based rating of health-related quality of life when used with clinical populations. For countries without a national value set, the EuroQol group recommends employing a value set based on geographic proximity. In the absence of a Swiss EQ-5D value set, we chose the French set. We used the EQ-5D version for adults and the EQ-5D version for youth.

In addition, patient's musculoskeletal function was evaluated using the 36-Item Short Form (SF-36, adult version) [16]. Results are typically used to calculate quality-adjusted life-years (QALYs) in the context of a cost-utility analysis. The SF-36 is comprised of questions covering eight domains: (1) physical functioning;(2) role limitations due to physical problems; (3) pain; (4) general health; (5) vitality; (6) social functioning; (7) role limitations due to emotional problems; and (8) mental health. Scores vary between 0 and 100 with higher scores indicating better musculoskeletal function and health-related quality of life.

Global evaluation of needs / bone treatment

A bone health evaluation was performed for all patients aged 5 years and older. All adults and children underwent a measurement of bone mineral density (BMD) by DXA (dual X-ray absorptiometry, Hologic Discovery A Hologic Inc., Bedford, MA), with measures at the spine and left proximal hip. We systematically added a vertebral fracture assessment based on DXA for all adults and children from 5 years of age. In adults, a trabecular bone score assessment completed the evaluation (TBS iNsight® Version 2.1 Med-Imaps, Pessac, France). The patient's bone metabolism was evaluated, including measurement of serum calcium, phosphorous, vitamin D, and bone turnover markers, and any anomaly was treated. Based on the clinical examination and bone evaluation results, an orthopaedic surgeon, a dentist and/or an otorhinolaryngologist was consulted for examinations and investigations. If necessary, patients were referred to other specialists for additional investigations (e.g., a cardiological assessment).

Access to genetic testing

All patients were offered a genetic consultation, analysis and counselling.

Synthesis of the initial evaluation

Results of this initial evaluation were discussed for each patient, during regular meetings of the OI team. A medical summary was provided to the primary care physician with the recommendations and proposals of the OI team, including medication.

Same follow up: the annual OI day, a patient- and family-centred day

After the initial evaluation, all patients were invited annually to a dedicated OI day organised in our clinic for an individualised medical check-up that was designed to address the patient's situation and needs (fig. 2).

The team of the OI day

For the OI day, the interdisciplinary team was reinforced by additional clinicians, DXA technicians (with two DXA machines), nurses and physiotherapists. The clinical space was expanded, and the team benefited from a designated meeting room and appropriate consultation rooms, with relevant available technology and equipment.

Evolution of the physical activity / evolution of the quality of live / evolution of the needs / bone treatment

Some of the examinations prescribed during the initial evaluation were repeated annually (physical examination, physiotherapy evaluation, blood analyses, EQ-5D, and SF-36), and others were done every 2 years (DXA) or as needed (orthopaedic surgeon, dentist and otorhinolaryngologist consultations). Transition consultations with both a paediatric and adult bone specialist were provided for teenagers (17 to 19 years old). A medical summary was provided to the primary care physician with the recommendations and proposals of the OI team, including medication.

Satisfaction of patients and their families:

On each OI day, a clinical and scientific information session was provided to patients, families and professional caregivers. This was intended to cover the latest advances in understanding and treatment, but also to connect people and to involve representatives of the national patients' association. To reach a broader audience, patients as well as representative of the national patients' association are invited to suggest a topic of interest for the next OI day. Satisfaction of patients and their families, restructuring of patients' psychosocial system and efficiency at providing a continuity of medical and surgical care were assessed by

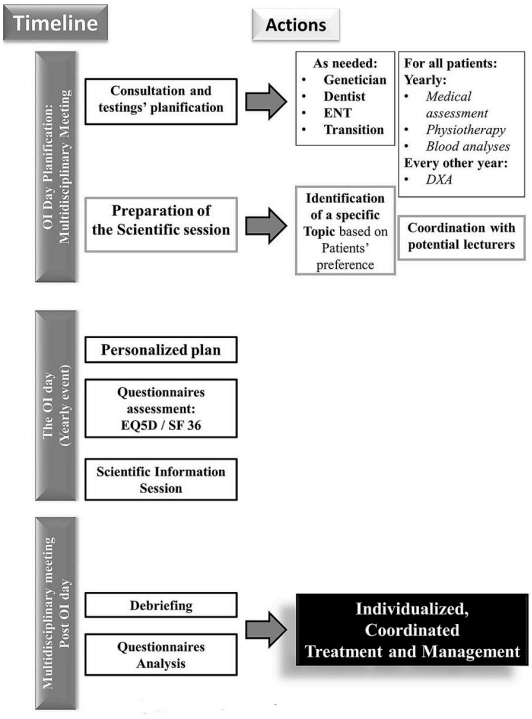


Figure 2: The OI day – planning of individualised follow-up. OI: Osteogenesis Imperfecta. CHUV Centre Hospitalier Universitaire Vaudois. DXA = dual X-ray absorptiometry.

measurement of the adherence to the OI day and the use of a short "OI day" quality survey (appendix 1). A typical timeline of an OI day for one patient is shown in figure S1 in appendix 2.

Statistical analysis

Statistical analyses were conducted using Stata ICv14® (StataCorp, College Station, Texas, USA) for Windows. Results were expressed as average ± standard deviation (SD). Association between studied parameters was determined using Student's t-test for continuous variables and chi-square test for categorical variables. Statistical significance was considered for a bivariate test with a p-value <0.05.

Results

Sample characteristics

Since 2012 we have met 71 new patients with OI, of whom 50 were included in this report. Exclusion criteria were: no consent agreement, loss of follow up (n = 20) or death (n = 1) (fig. 3). For the 50 patients included, the mean follow-up after the initial evaluation was 2.76 years (SD 1.57; minimum OI day = 1, maximum OI days = 5).

All the 50 patients included in this analysis received an initial evaluation. At the time of the first evaluation, 12 of these 50 patients were still of paediatric age (age 1 to 17 years, mean 8.5), and 38 were adults (age 18 to 69 years, mean 43.5) (table 1)

Figure 3: Patients excluded. OI = osteogenesis imperfecta

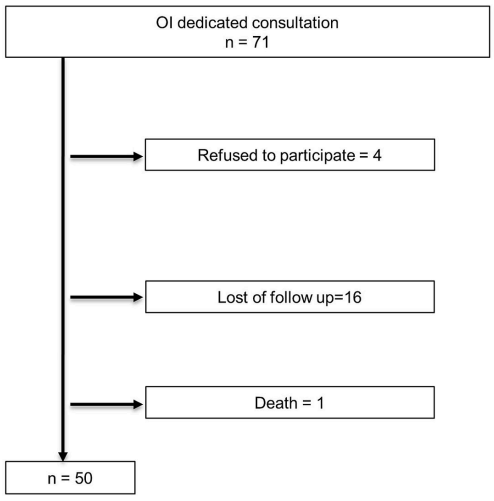


Table 1: Characteristics of the sample (n = 50).

Age (mean, min; max)	32.8 (1; 69)
Sex M/F (%)	22/68
Multiple previous fracture (%)	78
First fracture age (mean, min; max)	3.8 (0; 41)
Previous bone active drug (apart calcium / vitamin D) (%)	76

Physical activity: initial evaluation and evolution

We collected specific data on physical activity and sports for 45 patients (all except very young children). Initially, 24 of the 45 patients were not engaged in any type of specific physical activity (53%), whereas only 14 were participating in a sport (31%). Over time, 62% of the inactive patients started some physical activity and 44% started a sport. As of their last evaluation, the final rates of regular physical activity and sports were 82% and 60%, respectively, within the entire group.

Quality of life: initial evaluation and evolution

Thirty-six patients (72%; adults, children, teenagers) completed the EQ-5D at their initial evaluation, with an EQ-5D mean score of 0.75 (range 0.16 to 1.00; SD 0.17). However, we observed a tendency towards an improvement in the quality of life, with a mean score increasing from 0.73 to 0.75 (p >0.05) in the group of 26 patients with at least two EQ-5D measurements (mean time between two EQ-5D assessments 3.69 years). Thirty-four patients (68%; adults, teenagers) had an SF-36 evaluation at first evaluation, and 30 had at least two measurements. The initial SF-36 score, obtained during the initial evaluation (n = 34), demonstrated a lower score for the physical domain (mean 59.09; SD 4.15) than for the mental domain (mean 68.06; SD 3.66). Thirty patients presented with at least two SF-36 measurements (mean time between two SF-36 assessments 3.23 years). The physical domain score tended to increase between two assessment (from 59.09 to 65.79; p = 0.08), whereas it remained stable for the mental domain (score varying from 68.06 to 68.02).

Global evaluation of the needs / bone treatment: initial evaluation and evolution

About 80% (78%) of the whole cohort experienced multiple fractures in childhood; among these patients, 12 had never received any bone-active drug, apart from calcium / vitamin D substitution. All 38 adults, except one who had no measurable sites, had undergone at least one DXA measurement, with a mean spine T-score of −2.68 (range −5.6 to +0.6; SD 1.34), hip T-score −1.38 (range−3.3 to +1.6; SD 1.17), and neck T-score −1.59 (range −3.5 to +1.3; SD 1.25). Thirty-four adult patients underwent a bone texture measurement using a trabecular bone score method (TBS) with a mean spine TBS of 1.259 (range 1.003 to 1.501; SD 0.13, normal TBS >1.310). Only 5/12 children had a DXA measurement. Mean spine Z-score was −1.3 (range −1.9 to −1), hip Z-score was −2.0 (range −3.1 to −0.9), and neck Z-score −1.4 (only one measurement).

Genetic testing

Genetic assessment was obtained in 88% (38 adult patients) of our cohort patients. Eighteen patients from 11 families had a mutation in *COL1A1* predicting haploinsufficiency, associated with a mild phenotype. Four patients from three families had missense mutations in other amino acid residues of *COL1A1* or *COL1A2*, associated with a mild to moderate phenotype. A glycine substitution whether in *COL1A2* or in *COL1A1* (table 2, light grey), associated with a more severe phenotype, was observed in patients from six families. Both families with OI type V had the recurrent hotspot mutation in the 5′UTR region of *IFITM5*. One family with a dominant mild phenotype (type

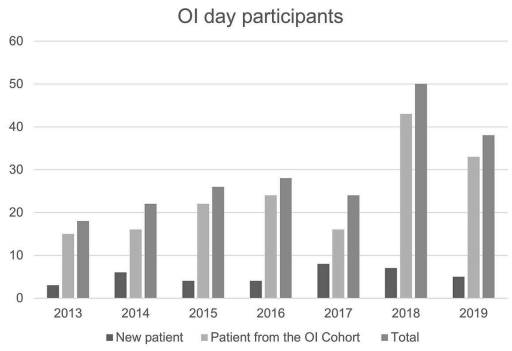
I) had a mutation in *CREB3L1*. The family with *LRP5* mutations included two affected patients: a heterozygous father with osteoporosis and a homozygous child with osteoporosis-pseudoglioma syndrome and a type I borderline to IV skeletal phenotype (table 2).

Satisfaction of patients and their families

Adherence to the OI day

Patients' attendance varied from year to year, from 27 patients in 2012 (creation of the OI day) to 50 in 2018 (seven new patients) and a slight decrease with 38 patients in 2019 (five new patients), this was later explained by a diminution in children's participation (fig. 4). From 2012 to 2018, the mean attendance to the OI days was 2.76 years (min 1 OI day, max 5 OI days).

Figure 4: Patient attendance at OI days. OI = osteogenesis imperfecta



OI day quality related survey

Thirty-five patients completed the survey, in an anonymous manner. The quality of the OI day was monitored through six main axes. Overall, most of the patients found the OI day useful for them (mean score=3.7 out of 4), improving their continuity of care (3.75 out of 4), their physical health (3.56 out of 4) (fig. 5). The impact of the OI day extended to the patient's family.

Discussion

Coordinated care in OI is sparse and limited. Development of a new interdisciplinary care strategy in a tertiary centre remains challenging and requires motivation driven towards a compelling purpose: patients' and their families' satisfaction. The enhanced teamwork provided by the interdisciplinary structure improved communication between the different stakeholders in OI patients' care. Thereby, it provided not only the ability to decide upon the best treatment plan for the patient, but it also brought sat-

Figure 5: OI day quality related survey. Patients answered with a grading scale: 1 = extremely dissatisfied, 2 = somewhat dissatisfied, 3 = somewhat satisfied, 4 = extremely satisfied.

Table 2: Phenotype and genotype of 38 patients from 24 families followed up from the osteogenesis imperfecta group.

Family (number of affected patients)	Skeletal phenotype	*Gene*	Mutation (cDNA)	Mutation (protein)
1 (1)	I	*COL1A1*	c.3536_3540delCCCCC	p.Pro1179ArgfsTer39
2 (2)	I	*COL1A1*	c.2426dupG	p.Ala811CysfsTer10
3 (4)	I	*COL1A1*	c.3540delC	p.Gly1181Alafs*58
4 (2)	I	*COL1A1*	c.2710delG	p.Glu904LysfsTer204
5 (1)	I	*COL1A1*	c.2867delG	p.Gly956AspfsTer152
6 (1)	I	*COL1A1*	c.1491delC	p.Ala498Glnfs*43
7 (1)	I	*COL1A1*	c.2614-1G>A	splicing
8 (1)	I	*COL1A1*	c.1477dupC	p.Arg493ProfsTer18
9 (2)	I	*COL1A1*	c.1405C>T	p.Arg469Ter
10 (1)	I	*COL1A1*	c.757C>T	p.Arg253Ter
11 (2)	I	*COL1A1*	c.658C>T	p.Arg220Ter
12 (1)	I	*COL1A2*	c.2797G>A	p.Asp933Asn
13 (1)	IV	*COL1A1*	c.3893C>A	p.Thr1298Asn
14 (2)	IV	*COL1A2*	c.3814T>C	p.Cys1272Arg
15 (2)	I-IV	*COL1A2*	c.964G>A	p.Gly322Ser
16 (1)	III	*COL1A2*	c.1378G>A	p.Gly460Ser
17 (1)	III	*COL1A2*	c.1678G>A	p.Gly560Ser
18 (1)	I-IV	*COL1A1*	c.581G>A	p.Gly194Asp
19 (1)	III	*COL1A2*	c.830G>A	p.Gly277Asp
20 (1)	III	*COL1A2*	c.1613G>T	p.Gly538Val
21 (3)	V	*IFITM5*	c.1-14C>T	Unknown
22 (1)	V	*IFITM5*	c.1-14C>T	Unknown
23 (3)	I	*CREB3L1*	c.p22C>T	p.Arg308Cys
24 (2)	I-IV	*LRP5*	c.1682C>T	p.Tyr581Ile

isfaction to the healthcare professionals, the patients and their families. Such impact of an interdisciplinary care strategy has been shown to foster faster [17] and better clinical decision making [18].

Our interdisciplinary team approach allowed us to offer a patient-centred clinical decision making, based on clinical observation, evidence-based guidelines and analyses of data collections. Regular data gathering provides support for optimised and comprehensive care strategies, but also allows the creation of data banks, which are extremely limited in OI. Our belief is that exhaustive data collection on a cohort that seems to be one of the largest in Europe can increase knowledge and improve patient care [9].

With the implementation of an interdisciplinary teamwork, we offered uniform but thorough evaluation and management for all patients.

All of the participants in this study had the same initial evaluation and participated in the OI day at least once. We evaluated the severity of the disease in order to propose the best treatment and management, using a classification based on the phenotype, the genotype, bone measurements (clinical and radiological) and a physiotherapeutic evaluation. Analyses of the phenotypes revealed a high number of patients with OI types I (58%), followed by type III (18%), and IV (14%). We met only four patients, from two different families, with OI type V. All patients were offered a genetic analysis, and 88% of them accepted. As expected, many of the mutations found were in *COL1A1* or *COL1A2*.

All adult patients but one (because of lack of any measurable site) underwent a complete bone evaluation. As expected, the mean bone mineral density values were in the range of bone fragility, and the mean TBS was low [19, 20]. The molecular findings, coupled with the DXA results, influenced the choice of bone therapy [4], leading to some modifications in all the patients' bone treatment in order to decrease their risk of fracture.

With the implementation of the interdisciplinary teamwork, we improved physical activity and the quality of life.

All of our patients had a physiotherapy evaluation and counselling in physical activities, 62% of inactive patients started some form of routine physical activity and 44% of patients not participating in any sports-related activity started to do so. Although most patients with mild to moderate OI can walk, they frequently report fatigue, diminished exercise capacity, kinesiophobia and exercise intolerance as limiting their regular physical and athletic activities, especially among children [21, 22]. In adult patients, physical activity was below population norms [23–25], even if patients were aware of their need to remain physically active to prevent loss of function and muscle strength. In one observational study, Wekre et al. identified a significant difference between the different types of OI and activity: patients with OI type III had less ability to perform activities than those with types I or IV [26]. In addition, Van Brussel et al. found that significant improvements in aerobic capacity and muscle force were observable after 3 months of training in children with OI. Unfortunately, these effects decreased over time as the intervention was discontinued [27]. As of their last evaluation, 82% of the CHUV OI group were actively participating in some form of routine physical activity and 60% were practicing sport. Among the nine patients with OI type III, only three were participating in at least one sport at their initial evaluation, but this number increased to six at the end of this study.

We discovered that our patients' quality of life was low in the physical health domain but was much better for mental health. We observed a clear but non-statistically significant trend toward an improved quality of life as reflected by an increase in EQ-5D scores. Although we lacked the statistical power to confirm a significant benefit over time, our results are nonetheless comparable with the literature. As already shown, the quality of life of OI patients is not severely impacted [28], except regarding physical function. Notably, their psychological and mental health is reported to be comparable to the healthy population [24, 25]. Having a coordinated organisation that includes assisting older adolescents transitioning from childhood to adulthood provides one of the best levels of care for children and adults [29]. Offering patients' group management, which helps to reduce their level of isolation, and personalised physical training has been shown to augment quality of life of OI children and their families [6, 27].

With implementation of the interdisciplinary team work through the OI day, we improved the satisfaction of patients and their families and helped to restructure patients' psychosocial systems

Patient satisfaction and restructuring of their psychosocial systems were measured using attendance at the OI day and its quality-related survey. Although participation varied from year to year, a large number of patients attended each year. For many of them, having all their medical evaluations conducted over the course of a single day was a real benefit in terms of organising, coordinating and simplifying their care. Regarding family and social participation, we originally proposed this OI day in order to address the problems of patient isolation and concerns about the financial burden of their care. In addition, we sought to facilitate the transition from adolescent to adult medical care for both the medical and physiotherapeutic components. This transition, acclaimed by families and caregivers, aimed to ensure continuity in medical management and to restructure psychosocial and work-related systems [29]. Even if we did not perform any economic analysis, the unique opportunity to have all of their evaluations and consultations on a single day once per year is unquestionably convenient and could explain the steady increase in new programme participants. Moreover, the plenary session seemed to be much appreciated by both patients and their families. They could meet other patients, share their experiences and keep in touch through social networking. The presence of a representative of the national patients association increased the association's visibility and established links with patients.

Programme strengths and weaknesses.

The main strengths of our interdisciplinary group were originally the OI day, which could answer to the majority of concerns, and the close collaboration with physical therapists. Physical activity and the quality of life improved.

The main limitation of our current programme pertains to the sustainability of the single OI day, given the increasing number of patients and the limited time and resources for organising the event. Even if managing rare diseases with an interdisciplinary approach is encouraged by our institution, reimbursement of medical care is comparable to other diseases despite the increased complexity of many cases. Moreover, the time spent in interdisciplinary meetings and organising the OI day is not recoverable. Reliable funding sources and better visibility would make this OI day sustainable on the long term. The second limitation related to confirming that interdisciplinary management improve the bone health and decrease the bone fragility: we have not enough data to date, but the OI day continues, and new dates are scheduled.

Conclusion

The interdisciplinary approach proposed by our group addressed most of the problems encountered by patients with OI. It satisfied the standard of care for improving the patients' condition, including their overall health and quality of life, in addition to familial and social participation [30–32]. It made it possible for patients to receive the clinical expertise and research-based wisdom of experts in their disease, resulting in optimised care [7].

This kind of management could help meeting rare disease patients' special needs. It provides a way to facilitate continuing education for team members and is consistent with the 2020 Rare Diseases Strategy of the Swiss Federal Council.

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Disclosure statement

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Appendix 1

OI day survey

Appendix 1 is available as a separate file at https://smw.ch/article/doi/smw.2020.20285.

Appendix 2

Supplementary figure

Figure S1: Timeline of an OI day, example for one patient. DXA = dual X-ray absorptiometry