Efficiency of a multidisciplinary approach to Osteogenesis Imperfecta: 6 years experience in a Swiss Tertiary Health Center.

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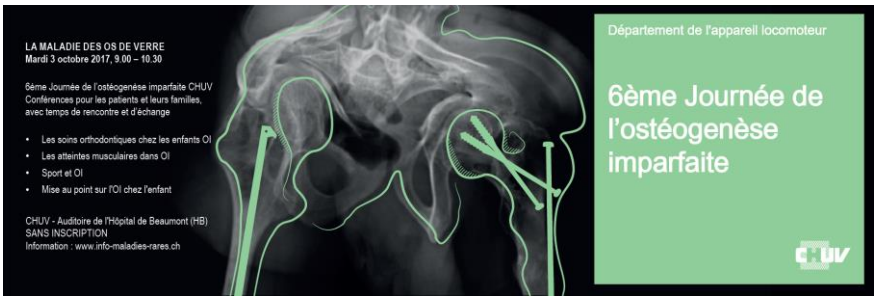
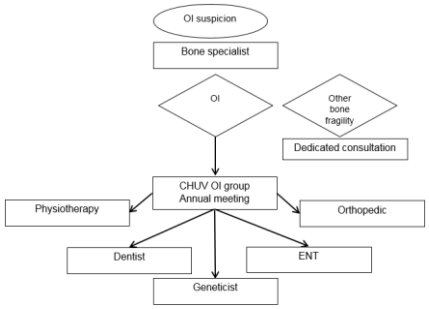
Introduction

Osteogenesis imperfecta (OI) is a rare genetic connective tissue disorder with wide phenotypic and molecular heterogeneity, causing risk of fractures in early life, progressive bone deformity, tooth and hearing alterations, and poor quality of life. In rare diseases, there is a real lack of patient information and recognition. Starting in 2012, we have employed a multidisciplinary approach for OI in our tertiary hospital, the Centre Hospitalier Universitaire Vaudois (CHUV), and created the CHUV OI group. The purpose of the present study is to evaluate the efficiency of this approach after 6 years.

Methods





An OI day is organized annually, and patients are invited to attend an individualized medical checkup, depending on their own situation and needs. The initial team was composed of two adult and pediatrician bone disorder specialists, an orthopedic surgeon, a geneticist and two physiotherapists (adult and pediatric). In 2013 and 2016, a dentist and a ENT specialist joined the team respectively. Each patient receives a physiotherapeutic evaluation with a proposition of physical therapy or counselling in physical activity and sport. In the same day, a clinical and scientific information session about the latest updates of the disease is organized, open to families and professional caregivers.

Results

50 patients have received a personalized medical evaluation since the beginning. 12 children (age1 to 17 years, mean 8.5) and 38 adults (age 18 to 69 years, mean 43.5) participated. The first year, 27 patients attended a medical check-up, 18 in 2013 (3 new patients), 22 in 2014 (6 new patients), 26 in 2015 (4 new patients), 28 in 2016 (4 new patients) and 24 in 2017 (8 new patients).
All adults, except 1 without any site measurable, had at least one DXA measurement, with a mean spine T score of -2.55 (-5.6; +0.6), hip T score -1.4 (-3.3; +1.6), neck T score -1.58 (-3.5; +1.3). 34 patients had a bone texture measurement by TBS with a mean spine TBS of 1.259 (1.003; 1.501).
Genetic evaluation was performed in 39 cases, and revealed mutation in 34 cases. One family has an osteoporosis pseudoglioma syndrome with LRP5 mutation; 2 families have OI type 5 with *IFITM5* mutations. Seven patients with OI type 3 have mutations in the type 1 collagen genes: 5 patients with a glycine substitution in *COL1A2,* 1 with a proline and 1 with an alanine substitution in *COL1A1*. Two additional patients had OI type 3 without genetic data. Four patients have OI type 4, but without mutation identified. The others have OI type 1: 3 with a glycine substitution in *COL1A2*, 1 with an aspartate substitution in *COL1A2*, and the remaining have haploinsufficiency mutations in *COL1A1*.
The majority of patients experienced multiple fractures in childhood, and 12 had never received any bone active drug, apart from calcium/vitamin D substitution. The multidisciplinary approach, including the DXA and genetic evaluation resulted in personalized treatment adaptation/new treatment/ for all patients.

Conclusion

The CHUV OI group multidisciplinary approach is efficient, resulting in better diagnosis, management and satisfaction of patients and their families as well as facilitating continuing education for the team members. Since 2012, the number of new patients has increased annually, with more patients benefiting of quality management including bone treatment and physical activity.

References

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