Abstract

**Purpose:** Skeletal dysplasias comprise a heterogenous group of genetic disorders that have generalized abnormalities in cartilage and bone. Although individually rare, collectively it is common with an estimate of 1 in 2000 to 3000. Individuals with skeletal dysplasias are known to be at risk for a myriad of medical complications associated with their conditions; hence a multidisciplinary approach to care is essential. This paper describes the opportunities and challenges in the creation of a multidisciplinary clinic for Filipino patients with skeletal dysplasia.

**Materials & methods:** A multidisciplinary clinic for Filipino patients with skeletal dysplasia was started. This is the first of its kind and made possible with the collaboration of the Departments of Pediatrics, Orthopedics and Rehabilitation Medicine of the Philippine General Hospital, in coordination with the Big Dreams for Little People – Philippines, an organization of Philippine-based people with dwarfism.

**Results:** A total of 25 patients in 21 families were seen in the clinic. 13 of the 25 patients (52%) were diagnosed clinically with achondroplasia. Medical concerns include hearing difficulty (3/25), low back pain (7/25), hip pain (2/25), ankle pain (1/25), and obstructive sleep apnea (1/25). Six of the 25 needed regular physical therapy.

**Conclusion:** Despite the limitations of molecular confirmation in many skeletal dysplasias and the challenges brought about by the Covid-19 pandemic, the multidisciplinary clinic for Filipino patients with skeletal dysplasia allowed awareness of patients and physicians of this group of disorders. In addition to better care directed towards the medical concerns of this special population, it is also hoped to improve health-seeking behavior of patients as access to healthcare is available.
Introduction

Skeletal dysplasias are a heterogenous group of hereditary disorders that have generalized abnormalities in cartilage and bone. Based on the recent nosology, there are more than 430 well-delineated disorders and more than 360 implicated genes in skeletal dysplasias [1]. Skeletal dysplasias, also known as osteochondrodysplasias, are individually rare but collectively common. Worldwide the estimated prevalence of skeletal dysplasias is between 2.1 to 4.7 per 10,000 [2].

This group of disorders can be diagnostically challenging. Rare disorders often are not clinically recognized early, hence, many of the medical concerns may also be neglected. As a primary diagnosis, skeletal dysplasias account for approximately 0.02% of hospitalizations for any condition in 2013 [3]. It is often that the primary diagnosis would reflect the problem associated with the condition, such as platyspondyly or lordosis, rather than the condition itself. This is related to the under-recognition of these conditions.

In the Philippines, access to this subspecialist level of care is limited to only a few tertiary hospitals. Medical geneticists and skeletal dysplasia specialists primarily handles these conditions. The medical geneticist to population density ratio in the Philippines is about 1:11,751,625 in 2013 [4]. In addition, these specialists are usually in the big cities and in the tertiary hospitals.

Due to the limited access of care, the under-recognition of this group of disorders, and the health-seeking behavior of people with skeletal dysplasia, a multidisciplinary clinic for these patients was established. This paper describes the opportunities and challenges noted after a multidisciplinary clinic for patients with skeletal dysplasia.

Materials and methods

The Section of Clinical Genetics under the Department of Pediatrics of University of the Philippines-Philippine General Hospital, initiated a multidisciplinary clinic for patients with dwarfism. This is in cooperation with the Departments of Orthopedics and Rehabilitation Medicine of the same institution and with the Big Dreams for Little People – Philippines, an organization of people with dwarfism which aims to create awareness, to advocate for the protection of the rights and welfare, and to promote equal opportunities for the little people in the Philippines.

A lay forum was initially held, attended by the patients and their families. Health education lectures about common skeletal dysplasia were tackled. A general overview of this group of disorders was discussed, including the mode of inheritance, clinical features, medical complications, and the medical, rehabilitation, and orthopedic interventions.

A separate half-day multispecialty clinic was organized. Informed consent was obtained from the patients before starting the clinic. The patients who attended were seen by the genetics, orthopedics and rehabilitation medicine services. These provided opportunities for our patients with skeletal dysplasia to seek consult for their medical concerns. For patients with no clinical diagnosis, skeletal surveys were facilitated and were followed-up at the outpatient clinic.

Results

A total of 25 patients in 21 families were seen in the clinic. Thirteen of the 25 patients (52%) were diagnosed clinically with achondroplasia, the most common skeletal dysplasia. Other cases seen include two patients with pseudoachondroplasia, one with hypochondroplasia, and one patient with brachytelephalangic chondrodysplasia punctata. Five patients had no definite diagnosis and required further evaluation through skeletal surveys. On follow-up, one patient was diagnosed with hypochondroplasia, one patient with acromesomelic dysplasia and another patient with multiple epiphyseal dysplasia based on the radiologic features. Two patients still need to follow-up. Medical concerns include hearing difficulty (3/25), low back pain (7/25), hip pain (2/25), ankle pain (1/25), and obstructive sleep apnea (1/25). Six of the 25 needed regular physical therapy.

Discussion

The optimal management of skeletal dysplasia requires a consideration of the medical, psychosocial and architectural consequences. Persons with skeletal dysplasia should be followed-up by a multidisciplinary team. There are patient-centered organizations which can be sources of information and support. These groups serve as good advocates for families and important partners in the care for the patients. In the Philippines, aside from the Big Dreams for Little People – Philippines, the Philippine Society for Orphan Disorders, can also be tapped. It is an organization which upholds the primary concerns and welfare of individuals with rare disorders by addressing, supporting and protecting their health and general well-being.

Clinical assessment alone may sometimes lead to the accurate diagnosis in some disorders like achondroplasia. Despite the significant advances in the genetics of skeletal dysplasia, radiographic skeletal survey remains to be the key diagnostic in most cases [5]. Hence, for patients whose clinical diagnosis is not apparent, skeletal survey was ordered.

Opportunities

The establishment of this multidisciplinary clinic emphasizes the complexity and the importance of healthcare in patients with skeletal dysplasia. With individual conditions being rare, most physicians are not adept at immediately recognizing these disorders including their complications. The diagnosis of skeletal dysplasia demands familiarity with numerous rare conditions and good pattern recognition skills [6] Having a multidisciplinary clinic helps physicians caring for these patients discuss and address concerns in the management with the other specialists. This allows the team of doctors to be able to reach to a diagnosis and therefore, treatment plan much faster and in a more cohesive manner. In Thailand, the Genetic Skeletal Dysplasia Clinic allowed education not only of the patients and their families, but also of the Thai pediatricians and orthopaedic surgeons [7].
At present, the frequency of this multidisciplinary clinic is every 6 months or twice a year. This is, however, complements the individual service’s clinics, which happens two to five times a week. With the increasing number of patients being seen and increasing awareness of physicians of this special clinic, it is foreseen that the frequency of holding this clinic will also be increased.

Through this multidisciplinary clinic, the patients become more aware of the availability of the services and their easy access. A definite diagnosis gives the patients some sense of control over their condition. With the knowledge of the specific diagnosis, physicians and patients monitor the complications more vigilantly. Awareness on the more common complications, skeletal and neurologic, makes compliance with treatment easier. Audiologic and ophthalmologic complications are also monitored. Psychosocial issues are also important concerns that need to be addressed. Hence, depending on the need, services and involvement in the management of other subspecialties are sourced to make management a holistic one. In the future, a needs assessment study may be done to identify additional services to be involved in the multidisciplinary clinic. Feedback from patients and their families may also be done to evaluate the clinic’s usefulness in the delivery of healthcare and service to patients with skeletal dysplasia.

Challenges

Although many of the disorders can be diagnosed clinically or by radiologic features, the importance of molecular confirmation has increasingly seen to be important. With the advances in the knowledge of the molecular basis of many skeletal dysplasias, the understanding of these conditions has been better. Hence, management options are becoming more specific and better directed. Also, establishment of a diagnosis is important in giving appropriate counselling regarding potential complications, expected adult height and recurrence risk [8].

Many of these molecular testing, at present, are still unavailable in the country and are expensive. Hence, this limitation may affect diagnosis and management of some skeletal dysplasias.

With the Covid–19 pandemic challenging healthcare systems all over the world, public health priority shifted towards response to the pandemic. Outpatient clinics, such as this, had been closed for a significant amount of time. At present, such service is still at a halt. But as seen in many settings, the utility of telemedicine may be explored.

The establishment of this multidisciplinary clinic is in response to and in accord with the World Health Organization’s Sustainable Development Goal 3: Ensure healthy lives and promote well-being for all at all ages [9]. This envisioned goal can be achieved by inclusion of patients with rare diseases, such as most skeletal dysplasias, in providing accessible quality essential healthcare services specific for them.

Conclusion

Despite the limitations of molecular confirmation in many skeletal dysplasias and the challenges brought about by the Covid–19 pandemic, the multidisciplinary clinic for Filipino patients with skeletal dysplasia allowed awareness of patients and physicians of this group of disorders. In addition to better care directed towards the medical concerns of this special population, it is also hoped to improve health-seeking behavior of patients as access to healthcare is available.

References


