The Kathryn O. and Alan C. Greenberg Center for Skeletal Dysplasias: An Interdisciplinary Approach

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Abstract Skeletal dysplasias are a group of over 300 genetic conditions often marked by short stature and a range of orthopedic problems. To meet the diverse medical, orthopedic, and psychosocial needs of individuals with skeletal dysplasias, the Kathryn O. and Alan C. Greenberg Center for Skeletal Dysplasias was organized at Hospital for Special Surgery in 2003. The center is the only one of its kind in the New York City metropolitan area and is dedicated to providing comprehensive medical care for individuals with skeletal dysplasias. The center is staffed by an interdisciplinary core team of health professionals consisting of an orthopedic surgeon, a medical geneticist, a genetic counselor/clinical coordinator, and a social worker. This interdisciplinary team of health professionals is committed to improving the quality of life for people with skeletal dysplasias through clinical care, research, education, and patient advocacy. Goals are achieved through a collaborative process that utilizes the expertise of the different professionals.

Keywords orthopedics · genetics · interdisciplinary care · skeletal dysplasia · dwarfism

Introduction

Over the past few decades, remarkable advances in medical technology, therapy, testing, and available treatments have changed health care delivery. Traditionally, health care services revolved around the primary care physician. He or she was responsible for recommending health care interventions and for referring patients to the appropriate specialist or specialists.

This multidisciplinary model has become increasingly complex and involves triaging care among disciplines that operate independently toward common patient goals. Members of multidisciplinary teams work separately, using the medical record as the chief means of communication.

Shared decision-making and flexible leadership characterizes interdisciplinary teamwork. First developed in the 1970s [1], the interdisciplinary team model serves as a medical communication system by which information exchange amongst the team of health care professionals has the end result of benefiting the patient. As a whole, the team has an identity that is separate from the identities of individual team members. Team members work collaboratively and meet regularly to discuss patient status and the evolving plan of care.

As traditional health care provider roles have become less-refined in pursuit of the most “cost-effective” health care, the use of the interdisciplinary model has declined [2]. In recent years, however, interest in interdisciplinary collaboration has risen in hospitals, community health care settings, and public health programs [3]. The skills of multiple disciplines working together in coordinated patient/family-focused care teams are necessary because patients and families have different vulnerabilities in front of different providers and each member of the team elicits different aspects of the patient’s and family’s illness experience.

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Established in 2003, the Kathryn O. and Alan C. Greenberg Center for Skeletal Dysplasias at Hospital for Special Surgery (HSS) uses an interdisciplinary approach to care for individuals and families affected by skeletal dysplasias. This paper presents some of our experiences with patients and families whose lives are affected by skeletal dysplasias.

Background

There are over 300 skeletal dysplasias, a heterogeneous group of genetic disorders characterized by differences in the size and shape of the limbs, trunk, and/or skull that often affect stature. Although individually rare, collectively there are a significant number of affected individuals. There is extreme variability among the clinical findings seen in the various dysplasias. Each skeletal dysplasia is classified by clinical findings, radiologic criteria, family history, and increasingly by genetic test results.

The Kathryn O. and Alan C. Greenberg Center for Skeletal Dysplasias' interdisciplinary approach allows continuity of care by a core team of specialists with experience of treating patients with these conditions. The members of the team, including a medical geneticist, orthopedic surgeon, clinical coordinator/genetic counselor, and a social worker, provide in-depth assessments and follow-up. Clinical services include genetic evaluation, diagnosis and counseling; orthopedic screening and treatment; psychosocial assessment; and referrals to a dedicated group of consultants for specialty medical evaluation/services. The interdisciplinary team develops an individualized treatment and management plan designed to meet the diagnostic, therapeutic, and long-term health needs of each individual with skeletal dysplasias and their families seeking services through the Center. This approach fosters continuity of care across a person's lifespan and bridges the pediatric–adulthood gap for individuals and families whose lives are affected by a skeletal dysplasia.

During the process of obtaining an appointment for their first visit at the Center, the clinical coordinator/genetic counselor obtains a brief history, intake information, and informs the patient/family about what to expect during their first visit to the center as well as the professionals they will meet during this evaluation. The clinical coordinator screens for medical and/or social issues which are of concern for each patient and communicates these concerns to the rest of the team. Appropriate arrangements are made for additional services, such as interpreters or wheelchair services inside the hospital, as needed during the patient's first visit.

During the first visit at the Center, each member of the core team meets with the patient and/or his/her family to obtain discipline-specific assessments. After the consultation is complete, the core Center staff discusses each patient at a scheduled case conference. The clinical coordinator/genetic counselor contacts the patient and/or family by letter and/or phone with the individualized follow-up plan and recommendations of the core team.

In addition, all patients seen through the Center are offered the option to enroll in a Skeletal Dysplasia Clinical Research Registry. The registry's goal is to gather information about each patient and their condition so that more information can be learned about the various dysplasias to develop future therapies/treatments for children and adults with skeletal dysplasias. Using the System for Collaborative Translational Research (SCTR), the Clinical Research Registry is currently used to collect clinical information and will gradually become a decision support tool, expanding to capture and analyze more data.

Chronic disease management programs should exist within a supportive, integrated, and comprehensive system of care in which the patient–provider relationship is central. Aston et al. examined interdisciplinary communication as an effective tool in enhancing communication and work processes among a team working in a pediatric specialty hospital. They concluded that interdisciplinary communication initiatives as simple as a regular discussion session can enhance personal and professional experience, leading to improved health outcomes for patients [4]. The chronic care model (CCM) aims to improve the care of patients with chronic illnesses, emphasizing six elements deemed to be essential for providing high-quality care to patients with chronic illnesses: delivery system design, self-management support, decision support, clinical information systems, community resources, and health care organization [5]. In a metaanalysis of empirical data from previously published literature on the topic, Tsai et al. determined that interventions utilizing at least one of the six chronic care model elements improve clinical outcomes, processes of care, and quality of life for patients with chronic illnesses. These results were consistent across a variety of chronic illnesses [6].

The quality of care and services the center provides have been supported by the feedback we have received from our patients. Patient satisfaction is a mainstay in health care today; however, in our efforts to use an interdisciplinary approach, it is paramount to have input from patients about their experiences as a member of the team.

Since its inception, the Center has actively surveyed patients and families for input regarding their initial and subsequent visits to the Center. A voluntary, confidential questionnaire was developed and administered to patients and/or their parents via telephone. All respondents were asked to answer nine questions concerning their first visit to the Center. Patients who returned to the Center for more than one visit were asked 11 additional questions. These questions focused on communication (staff-to-patient, interstaff) and environment (transportation to and from the hospital, lodging, getting around in the hospital, examination room accommodations) and included the option to include open-ended comments or suggestions regarding each question. Questions were also asked about follow-up services, referrals, and billing. Responses to all questions in each survey were given as a Likert scale with four options for patients to indicate their experiences: very good, good, fair, and poor.

Of the patient roster, 42% responded to the telephone survey (29/69). When answering questions about their first

visit to the Center, 22 of the surveyed patients (75.9%) ranked their comfort level with the overall center experience as “very good” (75.9%), and the remaining seven patients rated their experience as “good” (24.1%). Communication practices during the first Center visit also ranked positively amongst respondents. Twenty-four of the 29 respondents ranked communication between the Center staff and patient as “very good” (82.8%); the remaining five responded communication was “good” (17.2%). Twenty-three of the 29 respondents ranked communication amongst the staff as “very good” (79.3%), three respondents ranked it as “good” (10.3%), and three as “fair” (10.3%). One parent commented “The staff addressed my son, not me, which we both appreciated.”

Seventeen of the 29 respondents (58.6%) were eligible to complete the revisit questions. Twelve of the 17 revisit respondents indicated the highest rating of “very good” (70.6%) for their perception of being an active participant in their care, two respondents rated their own participation as “good” (11.8%), one as “fair” (5.9%), and one as “poor” (5.9%). Eleven of the 17 revisit respondents ranked follow-up visits as “very good” compared to their first visit (64.7%), five ranked follow-up visits as “good” (29.4%), and one respondent ranked follow-up as “fair” (5.9%).

Overall, the results of both the new patient and revisit survey indicate that patients seen at the Center are positive about the communication process offered by our interdisciplinary team and their participation as a member. Because the surveys are both anonymous and voluntary, it is difficult to establish specific factors affecting patient perception of events explored by the surveys. Future patient satisfaction surveys will need to be restructured to address these issues with more specificity.

Case vignettes

Case 1 Mucopolysaccharidosis type IH (MPS IH), also known as Hurler syndrome (OMIM 607014), is an autosomal recessive progressive lysosomal storage disorder caused by deficiency of the enzyme α -L-iduronidase (IDUA, EC 3.2.1.76) [7]. This enzyme is responsible for the normal degradation of the glycosaminoglycans (GAGs), heparan and dermatan sulfate, long chains of complex sugars with structural roles in the connective tissue, cartilage, and extracellular matrix of our tissues. Lysosomal accumulation of GAGs results in progressive organ dysfunction, widespread tissue damage, and physical and mental disabilities. Individuals with untreated Hurler syndrome present with progressive developmental delay, corneal clouding, airway obstructions, cardiac disease, hepatosplenomegaly, and dysostosis multiplex. Most die in the first decade of life from infections, aspiration, or cardiac failure. Bone marrow transplantation (BMT) performed before 24 months of age can ameliorate visceral and soft tissue manifestations, halt cognitive deterioration, and increase life expectancy, but has no effect on skeletal manifestations [8, 9].

We present a 14-year-old male who was diagnosed with Hurler syndrome at 10 months of age by enzymatic testing and subsequently underwent an allogenic bone marrow transplant at age 16 months. Surgical history includes corneal transplant, tonsillectomy and adenoidectomy, trigger digit releases, bilateral femoral and pelvic osteotomies, lumbar spine fusion 2002 (L1–L4), laminectomy with decompressive discectomy, and epiphyseal stapling of bilateral lower extremities for valgus deformity. Other pertinent history includes mitral valve prolapse, unilateral corneal clouding, and irritable bowel syndrome. He attends regular classes at school, and had participated on the basketball team. He presented with low back pain, bilateral hip pain which markedly increased over the prior 9 months, and marked right hip dysplasia. Until recently he ambulated on his own, but had been using a wheelchair and walker before surgery because of pain. The patient underwent a successful custom right total hip replacement with custom-cemented stem and acetabular cup. The following year, he underwent a successful left total hip replacement. Over the summer of 2007, he is working as a camp counselor and he will enter the 10th grade in fall.

The first successful bone marrow transplant in a patient with Hurler syndrome was completed in 1981. Previous studies have identified progressive hip deformity in these patients, in some cases requiring surgical containment procedures [7]. As this patient population ages beyond the life expectancy for untreated patients with Hurler syndrome, we expect an increased need for orthopedic intervention to preserve quality of life and independence in activities of daily living for these patients.

Case 2 Mucopolysaccharidosis type IV (MPS IV), also known as Morquio syndrome (OMIM 253000, 253010) is caused by a deficiency of the enzyme galactosamine-6-sulfatase (GALNS; EC 3.1.6.4) or β -galactosidase (EC 3.2.1.23) [10]. These enzymes are responsible for the normal degradation of the glycosaminoglycans (GAGs), keratan sulfate and chondroitin-6-sulfate, long chains of complex sugars with structural roles in the connective tissue, cartilage, and extracellular matrix of our tissues. Lysosomal accumulation of GAGs results in progressive organ dysfunction, widespread tissue damage, and physical disability. Individuals with Morquio syndrome typically have normal intelligence and present with a waddling gait, genu valgum, pes planus, and protruding sternum. Complications may arise from spinal cord compression secondary to atlantoaxial dislocation and/or acute thoracolumbar kyphosis. Most die in the second to third decade of life from cardiopulmonary failure. We present a 16-year-old girl who emigrated from China to New York with an undiagnosed bone disease. Her parents first noticed problems with her growth and the shape of her bones when she was 5 years old. Although they visited many specialists in China, no one was able to give them a diagnosis or information about ways to cure or alter her

condition. The family hoped to find a diagnosis and was in search of better care and improved quality of life for their daughter. Her condition had deteriorated to the point that she was no longer able to walk, and her parents had to carry her up and down the stairs to the walk-up apartment they shared with family.

Our geneticist brought them to the Kathryn O. and Alan C. Greenberg Center for Skeletal Dysplasias at HSS for evaluation and work-up. The patient and her family spoke no English, and we used a sensitive interpreter who worked closely with the family and medical team throughout the preoperative and postoperative process. This interpreter formed a bond with the family and continues to work with them at medical appointments. The center's staff worked with both the patient and her family to bridge cultural barriers between Chinese medicine where the family makes all decisions on behalf of the patient and Western medicine where the patient is involved in discussions regarding their medical and surgical care.

Through the Center's team approach, the patient and her family were given a diagnosis of Morquio syndrome, and it was determined that she needed surgical fusion of her cervical spine.

She underwent a successful cervical spinal fusion in 2004, and is now able to walk in her home. The Center's team assisted her family in applying for government-assisted housing, and her parents no longer have to carry her up and down the stairs to enter and leave the home. She now speaks English fluently, and will enter her senior year of high school next year. She attends her medical appointments and participates fully as an adult. Over the summer of 2007, she is completing an internship at HSS through the Center's Mentoring Program.

Case 3 Pseudoachondroplasia (PSACH; MIM 177170) is an autosomal dominant disorder caused by mutations in the gene for cartilage oligomeric matrix protein (COMP; MIM 600310) [11, 12]. The COMP protein is expressed in the extracellular matrix of the musculoskeletal tissues and is known to bind type II and IX collagen. Pseudoachondroplasia-causing mutations in COMP interfere with calcium-binding and affect the calcium-dependent folding process [13]. Studies in tissues with a pseudoachondroplasia COMP mutation have shown a decreased amount of COMP protein surrounding the cartilage cells (chondrocytes) in the growth plates of the bones. The COMP protein is instead compartmentalized within the cartilage cells inside the rough endoplasmic reticulum (rER). The build-up of COMP and other proteins (e.g., type IX Collagen) in the rER results in reduced and disorganized cartilage matrix in the growth plates of the bones as well as increased cartilage cell death. Thus, pseudoachondroplasia appears to be an endoplasmic reticulum storage disease, most likely caused by improper folding of COMP because of the pseudoachondroplasia mutation [11].

Individuals with pseudoachondroplasia present with decreased growth and waddling gait at 12–18 months of life.

Characteristic features include a disproportionately longer trunk with lumbar lordosis. Some individuals may have mild to moderate scoliosis. With the exception of the elbows, joints are hypermobile and the legs may be bowed or knock-kneed. Bones in the hands and feet tend to be shortened (brachydactyly). People with pseudoachondroplasia are generally of normal intelligence. Health problems commonly associated with pseudoachondroplasia include scoliosis, cervical spinal instability, and premature osteoarthritis. Individuals with pseudoachondroplasia are more likely to develop degenerative osteoarthritis at an earlier age than typical osteoarthritis, i.e., in the fourth to fifth decade [14].

We present a 47-year-old female who was diagnosed with pseudoachondroplasia at 18 months of age by clinical and radiological exam. Surgical history included femoral osteotomies at age 6 years and tibial osteotomies at age 12 years to correct genu varum, and correction of strabismus of the right eye at age 20 years. She had scoliosis and was treated with a Milwaukee brace at age 14 years. She presented to the Center with bilateral hip pain, left greater than right, and reported that she first began experiencing left hip pain at the age of 22 years. Although she did ambulate on her own, she had recently purchased a wheelchair because of the pain. The patient underwent a successful custom left total hip replacement with custom-cemented stem and acetabular cup.

Patients with pseudoachondroplasia often present at a relatively younger age with joint pain and restriction of movement. After conservative management fails to control these symptoms, lower extremity joint arthroplasty becomes an important surgical option for these patients [15].

Discussion

There is extreme variability among the various skeletal dysplasias, and they can be difficult to treat and manage. The interdisciplinary team approach used by the Kathryn O. and Alan C. Greenberg Center for Skeletal Dysplasias at HSS includes genetic evaluation, diagnosis, and counseling; orthopedic screening and treatment; psychosocial assessment; and referrals for specialty medical services. The core team develops an individualized treatment and management plan designed to meet the diagnostic, therapeutic, and long-term health needs of people with skeletal dysplasias and their families. This approach fosters continuity of care across a person's lifespan, and bridges the pediatric–adulthood gap for individuals and families whose lives are affected by a skeletal dysplasia.

The interdisciplinary team approach to care of patients with skeletal dysplasias has allowed the Center team to adopt a holistic approach, treating the whole individual, not simply their dysplasia. Continued efforts to maintain this methodology will allow for continuity of care across the lifespan for patients, ensuring efficient and safe patient care with the desired outcome of continued patient participation and satisfaction with their health care treatment.

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