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Perspective Article

Osteogenesis imperfecta: Effecting the transition from adolescent to adult medical care

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Abstract

The objective of this paper is to provide guidelines for pediatricians, adult physicians in different medical disciplines and patients' families who are planning the transition to adult care for the adolescent with osteogenesis imperfecta (OI). This observational report reflects concerns expressed by patients, their families, and involved physicians regarding the problems encountered with the transition of care. Methods for dealing with transitional issues are presented. OI is a heritable disorder of connective tissue in which fractures are the dominant clinical feature. However, OI is a systemic disorder with broad clinical variability in which there are unpredictable episodes of trauma. Coordinated team support provides the best level of care for the child with OI. This paper discusses 4 key topics related to effecting the transition from pediatric to adult care: 1) Transitioning and maintaining health, 2) Preserving or improving the level of function, 3) Assuring continuity of medical/surgical care, and 4) Re-structuring psychosocial and work-related systems. The process of transition requires active communication between the pediatric and adult team members along with a proactive approach by the patient and family. In addition, as the transition is established, the patient with OI should be encouraged to be his/her own advocate and care coordinator.

Keywords: Osteogenesis Imperfecta, Transiting Care, OI Child, OI Adult

Introduction

Transitional care is best defined by Philpott as "an organized effort to provide pediatric patients with the tools and resources they need to assure personal responsibility for their medical care while facilitating their transfer from a pediatrician to an adult practitioner"[1]. This effort has recently been addressed in the context of various chronic illnesses, e.g., inflammatory bowel disease, chronic neurologic diseases, and chronic cardiac disorders[2,3]. The transition to a new care system is particularly important for the young person with osteogenesis imperfecta (OI) in whom both comprehensive and readily accessible health care are important. Teaching the individual to assume responsibility for his/her medical care is essential when planning for transition. In OI this requires dealing with elements of care that are both acute and chronic. Two principles are important: first, competent immediate care is needed if there is a fracture. Ideally, a pediatric, and later, an adult orthopedic surgeon who knows the patient would be identified to consult regarding fractures. Second, adequate care in OI is dependent on team involvement, e.g., the family physician or internist, orthopedic surgeon, rehabilitation physiatrist, occupational therapist, nutritionist, as well as a social worker to deal with school and workplace issues. Physical therapy would be available as fracture healing is achieved. Severely affected OI patients may require supervision by a pulmonologist for restrictive lung disease caused by scoliosis. Effective transition requires involvement of the young adult with OI in the coordination of each of these disciplines which previously had been managed by the parents. In certain situations a formal team may not be available and instead the individual with OI must be proactive and become his/her own health coordinator by working with his pediatric physicians to find the appropriate future medical specialists, facilitate communication among the specialists, and obtain past medical records.

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Discussion

OI, a heritable disorder of connective tissue, is the most common of the inherited disorders primarily affecting bone. There are approximately 25,000 individuals with OI in the United States alone. It is estimated that twice that number are present, represented by individuals with mild OI in whom the diagnosis has not been made. Although the numbers of affected individuals is relatively small, this is a "high impact" disorder because of the unpredictable occurrence of fractures and the limited experience most physicians have in dealing with OI patients, either children or adults.

Fractures occurring with no injury or minor injury are the hallmark of OI. In children with OI, a fracture usually requires an unexpected trip to the Emergency Room, immobilization and/or surgery followed by sometimes long periods of rehabilitation. In addition to fractures, and depending on the severity of the OI, the patient with OI has to cope with restrictions on mobility which impact school and work. These factors and those addressed below, are critical issues as the adolescent outgrows the established pediatric care system and has to develop support within adult medicine, adult orthopedic surgery or other medical specialties. Experience indicates that this transition is anxiety-provoking for many patients (and their families) because of the difficulty encountered in finding an adult-oriented medical/social support system experienced in the care of adults with OI.

The adolescent with OI and his/her family face four key issues as they transition to the new adult care system: 1) maintaining their current state of health, 2) preserving or improving their level of function as physical demands expand, 3) assuring continuity of medical/surgical care, and 4) re-structuring psychosocial and work-related systems that previously were family-centric.

The transition phase is defined here as between 16 to 18 years of age. Planning should start approximately 2 years earlier.

Transition phase and the maintenance of current health status

When planning a new health care team the pediatrician/pediatric orthopedic surgeon should consider differences in care required by the mildly affected type I individual with OI versus those with more severe OI types (III-VII). Physician involvement with the patient with type I OI may be limited to periodic physical examinations aimed at surveying functional needs, while in the more severely involved patients, physician involvement should be more frequent, scheduled to assure compliance, and include such items as pulmonary assessment and correct wheelchair posture. It must be stressed that assembling an adequate team is difficult in adult medicine. Thus, it is the individual with OI who must become his/her own health care advocate managing overlap between the pediatric and adult team members.

Our experience is that many parents of adolescents with OI are not familiar with the scope of medical/surgical problems which may surface during this age. Subtle hearing loss may first occur in teenagers. We have encountered adolescents in whom aortic or mitral valve lesions were discovered for the first time requiring surgical replacement but for which the parents were unaware that this complication could surface at this age. Scoliosis may increase during the growth years requiring surgical correction. Approximately 30% of individuals with OI have dentinogenesis imperfecta which requires specialized dental care. In addition, adolescents and young adults with OI are at risk for developing renal calculi.

Although fractures, including vertebral fractures, are the dominant clinical feature, OI is a systemic connective tissue disorder with involvement of ligaments and tendons, muscle, the spine and peripheral joints, the eye, and the cardiovascular system. It is the potential for involvement of multiple organ systems with time that complicates the provision of adequate care for the OI patient.

It is of interest that while the frequency of fractures decreases after puberty, involvement of other connective tissue systems tends to increase with age. The ambulatory adolescent or college age student with mild or moderate type I OI is at risk for soft tissue injuries. Patients with excessive joint mobility may be at even greater risk. The young adult with a mild form of OI will want to participate in sports, and sports- related injuries are common in this age group, a matter which is of considerable concern to parents. Tendon and ligament injuries at the elbow and knees, such as anterior cruciate ligament tears, are representative of this type of injury which may require surgical repair[4]. For example, in this situation, if a gap in the transition to adult care occurs, insufficient attention to the initial injury may lead to a poor outcome. Moreover, achieving the cooperation of the young adult may pose a problem. Conflicting school schedules and physician appointments are problematic at this age. Finding competent medical care for complex injuries is a problem for the young adult at a time when the life situation is changing and extrinsic responsibilities are increasing.

Although the fracture risk falls after puberty, the wheel chair dependent adolescent or young adult with more severe OI (types III or IV) remains at great risk of accidental injury. In part, this is because their level of activity is increasing and more time is spent outside of the home. This is particularly the case as power or manual wheelchairs are used on city streets exposed to traffic. Teen-age drivers are at greater risk for accidents and automobile accidents are a significant cause of multiple fractures and long term disability in young patients with OI.

Preserving or improving their level of function

Change during the transition phase places considerable stress on the OI adolescents and their families. As individuals plan for college or work, a major concern is their ability to function in a new environment, particularly if distant from home. In college, the issues relate to scheduling classes consistent with the individual's level of function, finding suitable living quarters, and having the ability to get around campus buildings, particularly if in a wheelchair. OI patients frequently complain of decreased exercise tolerance, muscle weakness and fatigue. In responding to a question about muscle weakness, 37.7% of OI Registry respondents reported muscle weakness or discomfort and 28% reported experiencing muscle cramps. (Osteogenesis Imperfecta Registry, 2011). Recent studies of muscle fibers and responses to electrical stimulation in the *oim*/*oim* murine model of OI demonstrated that muscle fibers were smaller, contained less fibrillar collagen and had decreased peak titanic force response to electrical stimulation[5]. However, with appropriate training, muscle strength will improve and this is key to maintaining optimal function in the patient with OI. Our experience indicates that weight gain, which also limits mobility, is common in the adolescent with OI. Both nutritional counseling and appropriate exercise are important to maintain appropriate weight. Time constraints on the adolescent and family members may limit the utilization of physical therapy, but maintaining an active training program is critical to optimizing function as the child or young adult transitions to a new school or work environment. While impact sports are not encouraged, swimming, tennis, and hiking are good activities for ambulatory adolescents; swimming and vigorous upper extremity training and wheel-chair adapted activities are helpful in controlling weight in non-ambulatory adolescents and young adults.

Assuring continuity of medical/surgical care

Transferring to new medical care is a problem for most families dealing with OI, particularly those not living near major medical centers. This is because most physicians have had limited or no prior experience with young adults with OI. In the United States, insurance issues may also pose a problem when attempting to contact certain specialists. However, in this age of instant communication it should be possible for individuals or their physicians, even in remote communities, to obtain input from physicians experienced in the care of individuals with OI.

Re-structuring psychosocial and work-related systems

Here the major factors are: 1) educating the individual with OI to assume more responsibility for his/her medical care and 2) educating the individual with OI to identify potential barriers related to psychosocial, educational and work related issues.

Relationships and sexuality are concerns for the adolescent and young adult regardless of gender. It is important that the individual be counseled regarding reproductive issues prior to the age at which dating may occur. Even consideration of marriage should be discussed. Genetic counseling must be considered an important component of the individual's support network as marriage is considered.

In conclusion, what are the indicators of a successful transition from pediatric to adult care for an individual with OI in the 16-21 year age group? These would include factors that give the individual (and parents) the security in knowing that no gap would exist in the transfer of care and that a competent and committed medical/social care program had been achieved. A timely transfer of medical and surgical records between the pediatric and adult programs, including x-ray studies, is essential.

It is essential that the adult team members be identified and consulted as the transition is approached. The team includes the Family Physician, Orthopedist, Physiatrist or Physical Therapist, Occupational Therapist, Nutritionist, and Counselor or Social Worker.

System	**(%)**
Loss of vision	(12.6%)
Hearing loss	(19.6%)
Kidney Stones	(5.0%)
Diabetes	(1.9%)
Joint Dislocation	(24%)
Joint laxity	(55.6%)
Aortic or Mitral valve disease	(5.5%)
Scoliosis	(45.7%)
Fracture non-union	(9.3%)
High blood pressure	(5.3%)
Muscle weakness/cramps	(30.9%)

*System involvement as reported by 475 respondents ages 14-30 years in response to the osteogenesis imperfecta registry on-line questionnaire (2011)**
** Female: 306; male: 169*

Table 1. This table lists medical concerns identified by respondents in the adolescent to young adult age range to the questionnaire in the Osteogenesis Imperfecta Registry. Each of the disorders requires annual to periodic evaluation. The variety of disorders underscores the need to consider a coordinated approach to the provision of medical care in addition to that prompted by fractures.

In order for coordination to be achieved these team members should be in ready contact with each other and should have the necessary records in advance. The patient with OI, the primary care physician, and the orthopedist need to pre-establish a plan before acute intervention for injury is required. Trying to coordinate acute care and follow- up from the local emergency room will not work. Issues requiring periodic, if not annual, evaluations once the transition to adult care has been accomplished are listed in Table 1. The need for team cooperation is underscored by the inter-dependence of many of these medical concerns.

Consider the difficulties faced by an 18-year-old girl with type III OI and moderately severe scoliosis, who is wheel chair dependent, and searching for suitable freshman college dormitory facilities. She has been cared for by the same pediatric orthopedic group for many years. Traveling with her parents, during her initial weekend visit to a college campus, and at some distance from home, she suffers a humerus fracture requiring surgical repair. This scenario, which is not unusual for the OI family, points to the need for advance planning. A positive scenario would have included the following: being aware of her move to college and her home-based physicians and therapists, having already assisted the family by contacting the new health care team members at college-town, having previously discussed the level of care that would be needed, and having transferred records to the new team members. Thus, when the accident occurred proper care would be readily available. Not to be overlooked in the planning phase for this college student is consultation for non-fracture-related concerns such as contraception, her restrictive pulmonary function, and her chronic back pain related to scoliosis.

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