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Commentary

The Role of Physical Medicine and Rehabilitation in Managing Skeletal Dysplasia

Henry Wilson*

Department of Physiotherapy, University of Glasgow Caledonian, Glasgow, United Kingdom

DESCRIPTION

Skeletal dysplasia encompasses a group of more than 450 rare genetic disorders affecting the development, growth, and maintenance of the skeleton and connective tissue. These disorders present with diverse clinical features ranging from mild short stature to severe limb deformities and life-limiting complications. In Physical Medicine and Rehabilitation (PM&R), the role of the physiatrist becomes crucial in managing the functional consequences of skeletal dysplasia, aiming not only to alleviate pain and improve mobility but also to optimize independence, quality of life, and psychosocial well-being.

The clinical spectrum of skeletal dysplasias is broad. Conditions such as achondroplasia, osteogenesis imperfecta, spondyloepiphyseal dysplasia, and diastrophic dysplasia are among the more commonly recognized types, each with unique radiologic, genetic, and clinical hallmarks. Achondroplasia, for instance, is characterized by disproportionate short stature due to rhizomelic shortening, macrocephaly, and spinal stenosis, whereas osteogenesis imperfecta presents with increased bone fragility, blue sclerae, and frequent fractures. Despite the diversity in phenotype, individuals with skeletal dysplasia often share common rehabilitation challenges including joint contractures, muscle weakness, impaired gait, spinal deformities, and chronic pain.

From a rehabilitation perspective, the approach to skeletal dysplasia is inherently multidisciplinary. Physical medicine specialists must work closely with orthopedic surgeons, geneticists, pulmonologists, pain management experts, physical and occupational therapists, and psychologists. The comprehensive management plan begins with an individualized assessment that considers not only the specific dysplasia type but also the age, developmental stage, comorbidities, and personal goals of the patient. Functional limitations are assessed across domains such as mobility, Activities of Daily Living (ADLs), educational and vocational participation, and social integration.

For children with skeletal dysplasia, early intervention is key. Pediatric physiatrists emphasize developmental milestones, postural alignment, and joint integrity. Delayed gross motor milestones are common, requiring adaptive strategies and assistive technologies. In achondroplasia, for example, sitting and walking are often delayed due to hypotonia and macrocephaly. Physical therapy focuses on strengthening, flexibility, and safe mobility training. Occupational therapy assists with fine motor skills, adaptive self-care techniques, and modifications for school participation. The provision of appropriate mobility aids, such as walkers or wheelchairs, and environmental modifications can significantly enhance functional independence and prevent secondary complications.

Musculoskeletal pain and fatigue are recurring issues in patients with skeletal dysplasia. Pain can stem from mechanical stress due to joint misalignment, muscle imbalance, degenerative changes, or nerve impingement. Chronic back pain is particularly common in those with spinal deformities or stenosis. Physical medicine physicians employ a combination of pharmacological and non-pharmacological strategies for pain control. These include analgesics, muscle relaxants, activity modification, Transcutaneous Electrical Nerve Stimulation (TENS), and aquatic therapy. Cognitive behavioral therapy and mindfulness-based approaches are also integrated to address chronic pain and its psychological impact.

Rehabilitation also extends into the psychological and social domains. Living with a visible physical difference can pose significant psychosocial challenges. Children with skeletal dysplasia often experience bullying, low self-esteem, and social exclusion. Adults may face employment discrimination, relationship challenges, and accessibility barriers. The physiatrist addresses these concerns through supportive counseling, peer support networks, and community resource linkage. Social workers, vocational counselors, and mental health professionals are indispensable members of the rehabilitation team. Empowering patients through education, advocacy training, and assertiveness skills fosters autonomy and improves mental health outcomes.

Rehabilitation research in skeletal dysplasia, although growing, remains limited due to the rarity and heterogeneity of these

Correspondence to: Henry Wilson, Department of Physiotherapy, University of Glasgow Caledonian, Glasgow, United Kingdom, E-mail: wilsonh@gmail.com

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disorders. More studies are needed to establish evidence-based protocols for therapy intensity, assistive device selection, and surgical rehabilitation pathways. Patient-reported outcome measures specific to skeletal dysplasia are also underdeveloped, making it challenging to capture the full impact of interventions on daily life. However, emerging technologies such as motion analysis, virtual reality rehabilitation, and wearable sensors hold promise for advancing personalized rehabilitation.