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Journal of Osteoporosis and Physical Activity

Perspective

Idiopathic Osteoporosis: Causes, Diagnosis, Management, and Research Challenges in Young Adults

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DESCRIPTION

Idiopathic osteoporosis is a rare form of osteoporosis that occurs without any identifiable secondary cause, most often affecting young adults and premenopausal women. Unlike postmenopausal or steroid-induced osteoporosis, idiopathic osteoporosis arises in individuals who do not present with traditional risk factors such as hormonal deficiencies, chronic illnesses, or prolonged medication use. The term "idiopathic" reflects the unknown origin of the condition, which makes diagnosis and management particularly complex. Although it is less common than other types of osteoporosis, idiopathic osteoporosis can result in significant skeletal fragility, low Bone Mineral Density (BMD), and fractures that negatively impact quality of life.

This form of osteoporosis is typically divided into two main categories Juvenile Idiopathic Osteoporosis (JIO), which occurs in children and adolescents, and Adult Idiopathic Osteoporosis (AIO), which primarily affects young men and premenopausal women. Juvenile idiopathic osteoporosis usually appears before puberty and is often self-limiting, improving with the onset of puberty. In contrast, adult idiopathic osteoporosis tends to be persistent and may progress over time, leading to ongoing fracture risk.

The exact cause of idiopathic osteoporosis remains unknown, though several hypotheses have been proposed. Genetic factors are thought to play a major role. Individuals with idiopathic osteoporosis may have subtle abnormalities in genes involved in bone metabolism, such as those related to collagen production, vitamin D receptors, or bone remodeling enzymes. In some cases, there may be a family history of osteoporosis or fractures, suggesting a hereditary component. However, most patients do not present with any clear genetic mutation or known familial pattern.

Bone remodeling involves a balance between bone resorption by osteoclasts and bone formation by osteoblasts. In idiopathic osteoporosis, this balance is disrupted, leading to net bone loss. Some studies have suggested that individuals with idiopathic osteoporosis may have impaired osteoblast function or reduced bone formation rates. Others show increased bone turnover with an imbalance favoring resorption. However, these findings are not consistent across all patients, indicating that idiopathic osteoporosis is likely a heterogeneous condition with multiple contributing mechanisms.

Clinically, idiopathic osteoporosis is often first suspected after a low-trauma fracture, such as a vertebral compression fracture, hip fracture, or fracture of the wrist, especially in a young adult with no clear predisposing factors. Symptoms may include localized bone pain, back pain, height loss, or postural changes. In many cases, the condition remains undiagnosed until a fracture occurs, as routine screening for osteoporosis is not typically performed in younger adults without risk factors.

The diagnosis of idiopathic osteoporosis is made through a process of exclusion. A thorough medical history, physical examination, and laboratory tests are essential to rule out secondary causes of osteoporosis. These may include screening for hormonal imbalances (thyroid, parathyroid, cortisol levels), nutritional deficiencies (calcium, vitamin D), chronic inflammatory or gastrointestinal diseases (such as celiac disease or inflammatory bowel disease), and medication use (particularly glucocorticoids or anticonvulsants). Bone mineral density is measured using Dual-Energy X-ray Absorptiometry (DEXA), and a T-score of -2.5 or lower indicates osteoporosis. However, in premenopausal women and men under 50, Z-scores are more commonly used, with a Z-score below -2.0 considered below the expected range for age.

Bone turnover markers, such as serum osteocalcin, P1NP (Procollagen type 1 N-Terminal Propeptide), CTX (C-Terminal Telopeptide), and NTX (N-terminal telopeptide), may be measured to assess bone metabolic activity. Imaging studies, such as X-rays and MRI, help identify fractures or bone deformities and assess the severity of bone involvement.

Management of idiopathic osteoporosis focuses on preventing fractures, improving bone strength, and monitoring disease progression. Lifestyle modifications are crucial, including a

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Received: 20-Feb-2025, Manuscript No. JOPA-25-38090; **Editor assigned:** 24-Feb-2025, PreQC No. JOPA-25-38090 (PQ); **Reviewed:** 10-Mar-2025, QC No. JOPA-25-38090; **Revised:** 17-Mar-2025, Manuscript No. JOPA-25-38090 (R); **Published:** 24-Mar-2025, DOI: 10.35841/2329-9509.25.13.452

Citation: Sasaki T (2025). Idiopathic Osteoporosis: Causes, Diagnosis, Management, and Research Challenges in Young Adults. J Osteopor Phys Act.13:452.

balanced diet rich in calcium and vitamin D, weight-bearing and resistance exercises, and avoidance of smoking and excessive alcohol consumption. Calcium intake should reach 1,000-1,200 mg per day, and vitamin D supplementation should ensure adequate serum 25-hydroxyvitamin D levels above 30 ng/mL.

Pharmacologic treatment is considered for patients with documented low bone density and/or fragility fractures. Bisphosphonates are commonly used and have shown effectiveness in increasing BMD and reducing fracture risk in other types of osteoporosis. However, their use in premenopausal women and young adults remains somewhat controversial due to limited long-term safety data and potential effects on fertility and fetal development. Teriparatide, an anabolic agent that stimulates bone formation, may be considered in severe cases or when bisphosphonates are not effective. Denosumab is another option but also requires careful consideration due to potential rebound effects after discontinuation.

Because idiopathic osteoporosis often affects individuals during peak years of bone mass and physical activity, the psychosocial impact can be significant. Patients may experience anxiety, depression, or a sense of physical limitation due to fracture risk. Supportive care, education about the condition, and regular follow-up are essential components of comprehensive management.

Research into idiopathic osteoporosis is ongoing, and the condition remains a subject of clinical interest due to its unclear etiology and diverse presentations. Studies involving genetic profiling, bone biopsy, and advanced imaging may eventually help identify distinct subtypes and underlying mechanisms. Personalized treatment approaches based on individual bone metabolism profiles could improve outcomes and minimize unnecessary exposure to long-term medications.

CONCLUSION

Idiopathic osteoporosis is a rare but potentially debilitating bone disease that occurs in the absence of known secondary causes. It poses diagnostic and therapeutic challenges, especially in young individuals who would not typically be screened for osteoporosis. A combination of detailed clinical assessment, appropriate investigations, and individualized management strategies is necessary to reduce fracture risk and support long-term bone health in affected patients. Ongoing research is essential to uncover the causes and optimize treatment for this enigmatic condition.