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Commentary Article

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Current Laboratory Tools for Inherited Proteoglycan Biosynthesis Defects and Bikunin as a Promising Blood Biomarker

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Proteins are joined to sulfated glycosaminoglycan chains to form proteoglycans. They are a class of macromolecules that are primarily engaged in the construction of organs and tissues as key components of extracellular matrix. Some proteoglycans also function as signalling molecules in inflammatory responses, cell proliferation, adhesion, and differentiation. Inborn defects of proteoglycan metabolism are a group of orphan illnesses characterised by severe and permanent skeletal deformities as well as multiorgan dysfunction. Because of unspecific clinical symptoms, inaccessible functional laboratory tests, and a lack of simple blood biomarkers, identifying the gene variations that cause these diseases becomes challenging. We summarise the molecular routes of proteoglycan production, the accompanying hereditary disorders, and the relevant biochemical screening procedures in this review, with a specific emphasis on a circulating proteoglycan, termed bikunin, and its potential as a novel biomarker of these diseases.

Core proteins are connected to sulfated glycosaminoglycan (GAG) chains to form proteoglycans (PGs). They are a family of about fifty macromolecules that are involved in a wide range of pathophysiological processes in humans. During fetal development, growth, and aging, they transmit the biomechanical characteristics of the osteoarticular system as well as the connective tissues of nearly all organs. PGs are hydrophilic molecules found mostly in extracellular matrices (ECM), where they interact with one another as well as with hyaluronan and collagens. Such interactions are critical for the ECM's structure and viscoelastic characteristics. PGs also influence cell signalling by binding to a variety of ligands, including microbial pathogens, cytokines, and growth factors. As a result, they influence inflammatory and healing processes by encouraging cell recruitment and proliferation during infections, malignancies, and wound healing. The production of PG takes place in the endoplasmic reticulum (ER) and the Golgi apparatus, and it includes the interaction of multiple molecular actors throughout the secretory route. Inherited mutations that cause abnormalities in this system may lead to severe orphan illnesses known as "PG inherited metabolic disorders" (PGIMD). The causal variations are found in genes that encode enzymes involved for GAG chain elongation and modification. They might also have an impact on the proteins and transporters that govern the production and distribution of GAG assembly substrates. Osteoarticular malformations are common in PGIMD, and they are frequently accompanied with skin abnormalities, heart problems, neurological diseases, deafness, cataracts, and tooth abnormalities. Otherwise, mucopolysaccharidoses(MPS), a category of disorders caused by faulty GAG catabolism, exhibit clinical presentations that are strikingly similar to PGIMD. Pathogenic mutations in genes producing lysosomal GAGdegrading enzymes induce harmful GAG buildup in tissues. Given the very diverse and unspecific symptomatology of PGIMD, simple biochemical tests are required to direct attention to PGIMDcausing gene variations or to determine the causality of mutations revealed by whole exome sequencing. Whatever the method of diagnosis in PGIMD, contemporary laboratory techniques integrate genetic sequencing (gene panels or full exome sequencing) with PG content measurement in patientderived fibroblasts utilising substrate radiolabeling, chromatography, mass spectrometry (MS), and Western blotting. Bikunin (Bkn), a serum PG, has recently emerged as a possible novel biomarker for the fast identification of numerous PG biosynthetic defects. We give an overview of the structure of PGs, their biosynthesis routes, associated hereditary disorders, and current laboratory screening approaches in this paper. In addition, we explore the potentials and limitations of serum Bkn as a novel flexible biomarker for the screening and diagnosis of PGIMD.