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Ashyrova N.S., Geldimyradov Y.G.

DEVELOPMENT OF NEW INHIBITORS FOR FIBRODYSPLASIA OSSIFICANS PROGRESSIVA (FOP)

Tutor: senior lecture Podberezkina A.L.
Department of Biology
Belarusian State Medical University, Minsk

Relevance. Fibrodysplasia Ossificans Progressiva (FOP) is an extraordinarily rare genetic disorder, affecting approximately 1 in 2 million individuals worldwide. It causes soft tissues to progressively ossify into bone, leading to severe movement restrictions and disability. A key indicator of FOP is the malformation of the big toes observed at birth. The disorder results from a mutation in the ACVR1 gene-specifically the c.617G>A (p.R206H) mutation—which disrupts normal bone formation and causes uncontrolled ossification in muscles, tendons, and ligaments. Due to its rarity, FOP is often misdiagnosed as other conditions like arthritis, fibrous dysplasia, or cancer. Alarmingly, approximately 87% of patients undergo unnecessary surgical procedures, further worsening their condition. Early genetic testing plays a crucial role in ensuring accurate diagnosis and effective management.

Purpose: the aim of this study is to explore the effects of the ACVR1 mutation on abnormal ossification processes and its clinical implications, focusing on symptom progression, diagnosis accuracy, and potential management strategies to improve patient outcomes.

Materials and methods. The study examined the pathological role of the ACVR1 gene mutation in FOP patients, alongside their symptom progression and treatment responses. Data collection methods included imaging studies to identify ossification patterns and genetic testing to confirm mutations. Additionally, connections between the ACVR1 mutation and other pathologies, such as Diffuse Intrinsic Pontine Glioma (DIPG), were explored, with emphasis on shared BMP signaling pathway abnormalities.

Results and their discussion. Results indicate that the ACVR1 mutation contributes to uncontrolled activation of the BMP signaling pathway, leading to progressive ossification in FOP patients. Early genetic testing significantly improves diagnostic accuracy, preventing misdiagnoses and unnecessary interventions.

Treatment approaches reduce inflammation and swelling during flare-ups, enhancing patient comfort. However, physical trauma, even minor injuries, triggers new ossification events, making preventive care critical. The study highlights the importance of patient-specific management plans to mitigate complications and preserve mobility.

The analysis also reveals that the ACVR1 mutation plays a role in DIPG, though the mutation timeline differs between the two conditions. Both diseases involve excessive tissue growth driven by BMP pathway dysregulation, offering a promising direction for targeted therapies.

Conclusions. Diagnostic Precision: Early genetic testing is vital for accurate diagnosis and preventing unnecessary treatments. Effective Management: Medication and preventive measures improve quality of life for FOP patients while mitigating disease progression.