THE FIRST IDENTIFIED CASE OF HEREDITARY ANGIOEDEMA IN KOSOVA

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At a time when diagnosing rare diseases in Kosovo remains a continuous challenge, cases like this represent not only an individual success but also an important step toward building a more sensitive and informed healthcare system. Dr. Rushit Ismajli, a specialist in family medicine in Prishtina, identified the first documented case of Hereditary Angioedema (HAE) in Kosovo – an achievement presented internationally at the Family Medicine Conference in Munich, Germany (2023). This case not only set a scientific precedent but also led to a practical result: the inclusion of the drug C1 esterase inhibitor in Kosovo's Essential Medicines List, thanks to the dedication and collaboration of Dr. Shqipe Spahiu Konjusha, a rare disease expert at the University Clinical Center of Kosovo (UCCK).

This publication aims to raise awareness within the medical community about the importance of early diagnosis of rare diseases, the role of family doctors in this process, and the necessity of inter-institutional and regional cooperation to ensure appropriate treatment. The reported case demonstrates the power of knowledge, clinical curiosity, and professional commitment in improving the quality of life of patients in Kosovo.

Introduction

Hereditary Angioedema (HAE) is a rare, potentially life-threatening genetic disorder affecting about 1 in 10,000 to 1 in 50,000 individuals, with no significant differences among ethnic groups. In Kosovo, until 2023, HAE had not been officially reported. The presented case represents the first confirmed case of HAE in the country, marking a key moment for the diagnosis and management of rare diseases in family medicine practice.

Case Presentation

The patient E.S., a 23-year-old male, had experienced recurrent episodes of unexplained swelling in the hands, feet, lips, and genital organs over several years. He twice developed airway swelling, posing a risk of asphyxiation. Initially, his symptoms were treated with corticosteroids and antihistamines, under the assumption of allergic reactions. He had also undergone an appendectomy (now believed to be related to HAE).

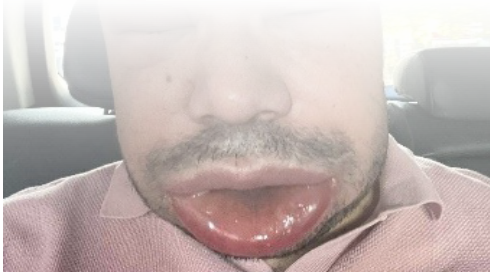

Figure 1. A.M. 36 years old, during the face attack.

Due to the lack of local awareness about HAE, the reporting physician sought information from international sources and contacted the patient's relatives, who reported similar symptoms among family members living in Switzerland. After consultation with Natasha Ageleska, Regional Patient Advocate for HAE in Southeastern Europe, the patient was referred to North Macedonia, where laboratory analyses confirmed the diagnosis:

- C1q complement: 30.3 (normal range 13.0–32.0)

- C4 complement: 14.6 (normal range 12–36)

- C1 esterase inhibitor: 40 (normal range 70–130)

Final diagnosis: Hereditary Angioedema due to C1 esterase inhibitor deficiency.

Diagnostic Challenges and Cross-Border Collaboration

In the absence of disease recognition within Kosovo, both the patient and the family doctor faced major difficulties in securing diagnosis and treatment. Assistance from colleagues in North Macedonia was crucial in confirming the laboratory results and guiding the patient toward appropriate therapy.

Improvement of the Situation in Kosovo

Through cooperation with Dr. Shqipe Spahiu Konjusha, pediatric geneticist and coordinator for rare diseases at UCCK, C1 esterase inhibitor was added to Kosovo's Essential Medicines List, supported by the Ministry of Health. This step made HAE treatment accessible to patients in Kosovo and represents a major achievement in rare disease care. Meanwhile, several other patients have been identified and are being treated at the Allergology Clinic of UCCK.

Discussion

The case underscores the critical role of the family doctor in the early identification of rare diseases and the importance of regional and international cooperation. Educating healthcare personnel to distinguish HAE from allergic reactions is essential to prevent fatal outcomes.

Conclusion

The identification of this case marks a historic milestone for Kosovar medicine. It demonstrates

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that collaboration, clinical curiosity, and professional dedication can lead to real improvement in healthcare. The inclusion of C1 esterase inhibitor in the Essential Medicines List represents a victory for patients with rare diseases and a model of successful synergy between clinical practice, health policy, and the medical community.

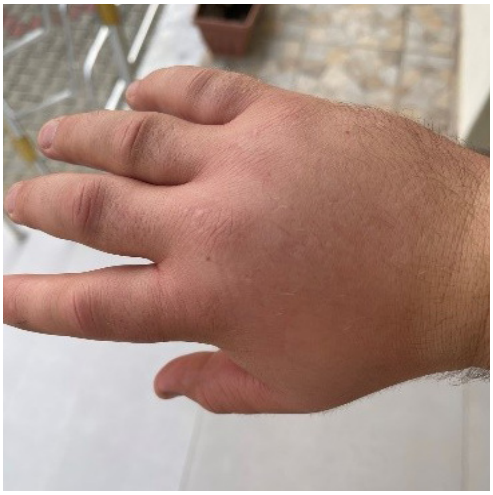

Figure 2. E.S. 25 years old, during the hand attack

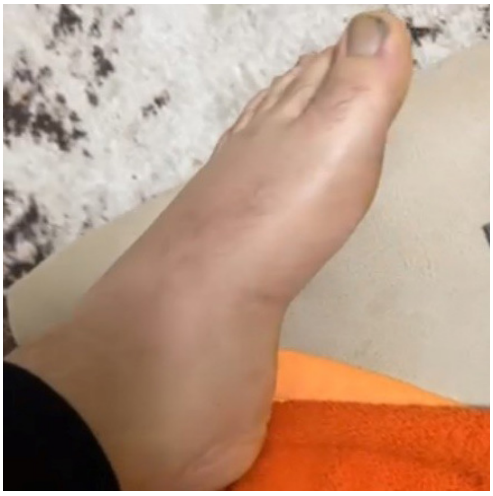

Figure 3. E.S. 25 years old, during the foot attack.

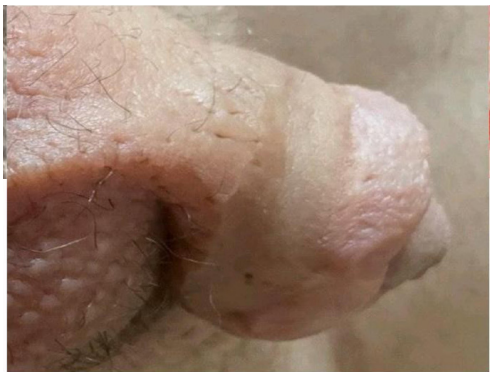

Figure 4. E.S. 25 years old, during the scrotal attack.

Referencat:

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