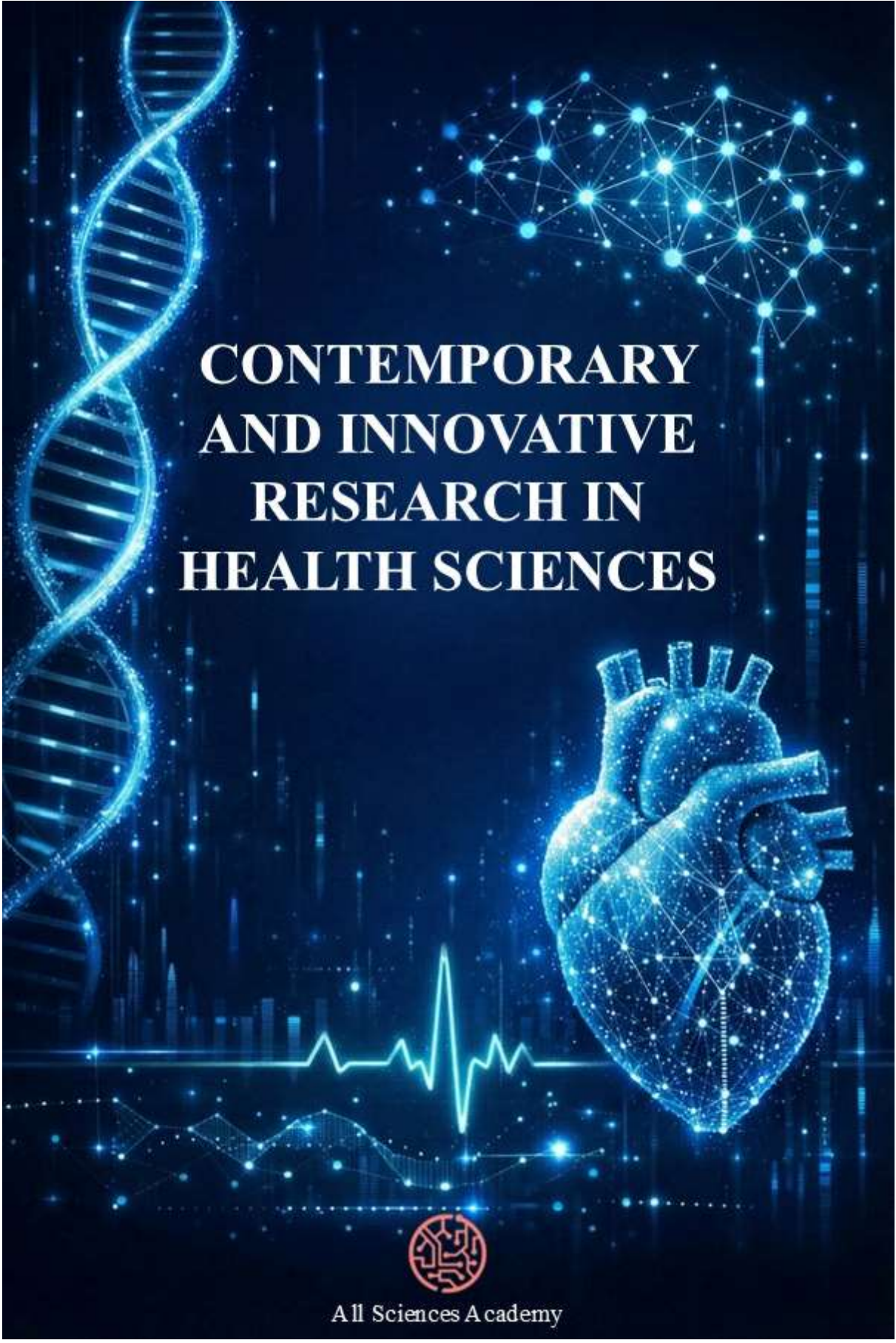
CONTEMPORARY AND INNOVATIVE RESEARCH IN HEALTH SCIENCES



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CONTENT

A Rare Racopathy in Childhood: Costello Syndrome Case Report and Nursing Care

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ABSTRACT

Costello syndrome is a rare developmental disorder associated with HRAS gene mutation, affecting multiple systems. In this syndrome, serious clinical problems such as growth and developmental retardation, feeding difficulties, hypotonia, cardiac anomalies and increased tumor risk are seen. Focusing only on medical treatment in children with Costello syndrome is insufficient to improve the long-term consequences of the disease; the absence of systematic and continuous nursing care can lead to a rapid deterioration of the child's clinical condition and a significant decrease in the quality of life. In cases where nursing care is not adequately planned, nutritional deficiencies deepen, growth-developmental retardation progresses, the risk of infection increases and complications due to cardiac problems are more common. In addition, the non-implementation of appropriate nursing initiatives for hypotonia and motor developmental delay negatively affects the level of independence and functional capacity of the child. The fact that families are not sufficiently supported in the care process increases the burden of care and reduces compliance with treatment. In this context, nursing care in Costello syndrome is critical in terms of meeting the basic needs of the child, noticing and preventing complications at an early stage, closely monitoring growth and development, and ensuring the active participation of the care process by educating the family. Planned, individualized and evidence-based nursing care improves the quality of life of children with Costello syndrome and positively affects the long-term outcomes of the disease. The purpose of this book section is to emphasize the indispensable role of nursing care in Costello syndrome and to provide a care approach that will guide clinical practices.

Keywords: Costello Syndrome, Genetic Diseases, Pediatric Nursing, Nursing Care, With Care Diseases.

INTRODUCTION

Costello syndrome is a rare genetic disorder resulting from pathogenic variants in the *HRAS* gene (Gripp & Rauen, 2024). It is classified within the group of RASopathies, a heterogeneous spectrum of developmental disorders characterized by a shared molecular etiology involving dysregulation of the RAS/mitogen-activated protein kinase (RAS/MAPK) signaling pathway.

The core clinical phenotype encompasses postnatal growth failure, global developmental delay, intellectual disability, coarse facial features, and significant feeding difficulties (StatPearls, 2025). In infancy, the condition is typically marked by generalized hypotonia, severe feeding impairment, short stature, cognitive deficits, distinctive craniofacial characteristics, curly or sparse hair, soft and hyperextensible skin, papillomata, joint laxity, and cardiac involvement.

Furthermore, postnatal cerebellar overgrowth has been reported and may predispose affected individuals to hydrocephalus or syringomyelia in association with Chiari type I malformation. The lifetime cumulative risk of malignancy is estimated at approximately 15%. The most frequently reported neoplasms include rhabdomyosarcoma and neuroblastoma during childhood, as well as transitional cell carcinoma of the bladder in adolescence and early adulthood (RASopathies Network, 2025).

Costello syndrome is an exceedingly rare disorder worldwide. The estimated birth prevalence is approximately 1 in 1,380,000 in the United Kingdom and 1 in 1,230,000 in Japan (Gripp & Rauen, 2024). These variations are likely attributable to phenotypic heterogeneity, diagnostic challenges, and potential misclassification with other RASopathies. Owing to its rarity, broad clinical spectrum, and substantial cardiac and oncologic risks, early diagnosis and structured multidisciplinary follow-up are of critical importance (Leoni et al., 2022).

In rare, multisystem genetic disorders such as Costello syndrome, nursing care extends beyond symptom monitoring and implementation of clinical interventions, necessitating comprehensive, long-term, and systematic follow-up (Leoni et al., 2022). Early recognition of complications and maintenance of continuity of care constitute fundamental objectives of nursing management in order to enhance the quality of life of affected individuals (StatPearls, 2025). Within this framework, the importance of a multidisciplinary approach is emphasized, and nurses assume a pivotal role in coordinating care between the patient, family, and healthcare team. Their responsibilities include monitoring growth and development, managing feeding difficulties, and conducting regular surveillance of cardiac and neurological manifestations (Gripp & Rauen, 2024).

The high prevalence of hypotonia, feeding disorders, and developmental delays during infancy and early childhood further underscores the necessity of thorough nursing assessments and individualized care plans. Moreover, given the lifelong increased risk of malignancy and potential cardiac complications, family education and counseling regarding possible risks are integral components of nursing care (Leoni et al., 2022). Nursing management in Costello

syndrome plays a vital role in improving health outcomes and quality of life in pediatric populations. This report aims to contribute to the limited literature concerning the clinical characteristics of Costello syndrome in childhood and to emphasize the significance of nursing care in its management.

Pathophysiology of Costello Syndrome

Costello syndrome is caused by activating mutations in the *HRAS* gene, which result in constitutive upregulation of the RAS/mitogen-activated protein kinase (RAS/MAPK) signaling pathway. Sustained pathway activation disrupts the physiological regulation of cellular proliferation, differentiation, and apoptosis. This molecular dysregulation leads to aberrant intracellular signaling, altered growth factor responses, and impaired tissue homeostasis. Consequently, a wide clinical spectrum emerges, encompassing cardiac hypertrophy and arrhythmogenesis, connective tissue abnormalities, decreased dermal elasticity, and a heightened predisposition to neoplastic transformation (Gripp & Rauen, 2024). Clinical features are typically evident from birth. Common findings include macrocephaly, coarse facial appearance, redundant or lax skin, papillomata, postnatal growth failure, and global developmental delay (StatPearls, 2025).

Neurological involvement is frequently observed and includes generalized hypotonia, epileptic seizures, structural brain anomalies, and behavioral characteristics resembling autism spectrum disorder (Leoni et al., 2022).

Cardiovascular involvement is highly prevalent and represents a major contributor to morbidity and mortality. Hypertrophic cardiomyopathy, congenital heart defects, and atrial and/or ventricular arrhythmias significantly increase the risk of adverse clinical outcomes (Gripp & Rauen, 2024; Uysal et al., 2013).

Furthermore, individuals with Costello syndrome have an increased lifetime risk of malignancies, particularly rhabdomyosarcoma, neuroblastoma, and transitional cell carcinoma of the bladder (StatPearls, 2025; Failoc-Rojas et al., 2023).

Diagnosis of Costello Syndrome

Diagnosis begins with clinical evaluation and analysis of phenotypic features. Definitive diagnosis is established by demonstrating a mutation in the HRAS gene through genetic testing. Costello syndrome is an autosomal dominant

RASopathy, and molecular confirmation represents the gold standard for diagnosis.

For cardiac assessment, echocardiography and electrocardiography are recommended. Neurological and psychometric tests are used to evaluate developmental status. Due to the increased risk of malignancy, abdominal ultrasonography and magnetic resonance imaging are also recommended (Gripp & Rauen, 2024).

Treatment is symptomatic and supportive. Management of cardiac complications, developmental support programs, treatment of skin lesions, and surgical interventions for orthopedic problems constitute the main approaches (Grabala et al., 2024).

Because of the increased malignancy risk, regular oncological screenings should be performed, and follow-up should be maintained by a multidisciplinary team. Genetic counseling is also important for informing families about the disease course, recurrence risk, and prenatal diagnostic options

Case Scenario

This case, describing the care and clinical course of a child diagnosed with Costello syndrome, was prepared based on current literature data (Grabala et al., 2024; Gripp & Rauen, 2024; Perrino et al., 2024; Zenker, 2022).

Six-year-old A.E. was brought to the hospital by his family due to developmental delay, loss of speech, and neurological symptoms. He was born at 36+4 weeks of gestation via cesarean section and was followed in the Neonatal Intensive Care Unit (NICU) for four months during the postnatal period. During his NICU stay, a cardiac arrhythmia was detected, and propranolol (Dideral) therapy was initiated.

Upon review of his developmental history, it was learned that he was able to use single words at 18 months of age; however, he later lost his acquired language skills. His vaccinations were reported to be up to date. His height was measured as 108 cm and his weight as 15 kg. He completed toilet training at 4.5 years of age. Marked weakness in fine motor skills and hand muscle strength was noted. He had significant chewing and swallowing difficulties. Due to problems with social adaptation, he was receiving special education services and was reported to exhibit autistic features.

Neurological evaluation revealed that he had been diagnosed with epilepsy due to nocturnal seizure episodes and was receiving levetiracetam (Keppra) treatment. His secondary diagnoses included epilepsy, microcephaly, and autism spectrum disorder.

Evaluation of the family history showed no consanguinity between the parents, and both were described as educated and well-informed. The patient's first siblings, twin sisters, were also under developmental follow-up. Both had speech delay, incomplete word production, and a diagnosis of epilepsy; they were receiving valproic acid (Depakin) treatment, and one of the twins had additionally been diagnosed with autism spectrum disorder. In light of all these findings, genetic analysis revealed a mutation in the HRAS gene, and the patient was diagnosed with Costello syndrome.

Nursing Approaches and Care Plan in Costello syndrome

Treatment is symptomatic and supportive in nature. Management of cardiac complications, implementation of developmental support programs, treatment of skin lesions, and surgical interventions for orthopedic problems constitute the main approaches. Due to the increased risk of malignancy, regular oncological screenings should be performed, and follow-up should be maintained by a multidisciplinary team (Grabala et al., 2024). Genetic counseling is also essential to increase families' awareness of the disease process and its implications.

Nursing care plans are systematically developed using NANDA-I nursing diagnoses, NIC interventions, and NOC outcomes, and these structured systems influence all stages of the nursing process. This approach ensures that nursing care is holistic, patient-centered, and delivered in a standardized yet individualized manner. Through these methods, diagnoses, interventions, and outcomes in the care process are clearly planned, implemented, and evaluated (NANDA International, 2024; Hockenberry & Wilson, 2023).

Tablo 1.Costello Sendromuna İlişkin Hemşirelik Tanıları ve Bakım Planı

NANDA NURSING DIAGNOSIS	**CLINICAL INDICATORS**	**EXPECTED OUTCOMES (NOC)**	**NURSING INTERVENTIONS (NIC)**	**EVALUATION**
1. Yutmada bozulma → Impaired Swallowing	• Impaired coordination of chewing and swallowing • OOropharyngeal muscle weakness	To reduce the risk of aspiration and ensure safe oral intake.	• The patient was positioned appropriately (semi-Fowler's position) during feeding. • The consistency of foods was modified/thinned. (Perrino et al., 2024) • The patient was monitored for signs of aspiration (coughing, cyanosis, respiratory distress)..	• Tolerance to oral intake increased. • No cyanosis or signs of aspiration were observed during feeding.

2. Aspirasyon riski → Risk for Aspiration	• Decreased swallowing reflex*)*	Prevention of aspiration.	• Respiratory status was closely monitored during feeding. • An appropriate feeding position was maintained. (Perrino et al., 2024)	• No evidence of aspiration was observed.

Tablo 1.Costello Sendromuna İlişkin Hemşirelik Tanıları ve Bakım Planı (Devamı)

NANDA NURSING DIAGNOSIS	**CLINICAL INDICATORS**	**EXPECTED OUTCOMES (NOC)**	**NURSING INTERVENTIONS (NIC)**	**EVALUATION**
3. Imbalanced Nutrition: Less Than Body Requirements	Impaired Swallowing and Increased Metabolic Demand	Achievement of adequate nutrition and caloric intake.	• Daily dietary intake was monitored. (Perrino et al., 2024) • A high-calorie nutrition plan was developed. (Perrino et al., 2024)	Weight and growth parameters remained stable.

4. Impaired Physical Mobility	Decreased Muscle Tone and Delayed Fine Motor Skills	Enhancement of motor function and muscle power	• Age-appropriate daily exercises were encouraged. • Collaboration with the physiotherapy team was ensured. (Perrino et al., 2024) • Assistance was provided during physical activities. (Perrino et al., 2024)	An improvement in fine motor skills and muscle strength was observed.

NANDA NURSING DIAGNOSIS	CLINICAL INDICATORS	EXPECTED OUTCOMES (NOC)	NURSING INTERVENTIONS (NIC)	EVALUATION
5. Delayed Growth and Development	Costello syndrome and history of prematurity	Support of developmental milestones	• Developmental assessments were performed. (Perrino et al., 2024) • The family was educated on developmentally supportive care. (Perrino et al., 2024)	Progress was observed in developmental milestones.
6. Sosyal Impaired Social Interaction	Adaptation difficulties, limited communication	Enhancement of social interaction	• Participation in play and group activities was encouraged. (Perrino et al., 2024) • Positive behaviors were reinforced.	An increase was noted in social interaction initiatives.

7. Impaired Verbal Communication	Developmental delay, neuromotor impairment	Promotion of communication skills	• Simple, clear, and age-appropriate communication techniques were used. Visual aids and gestures were employed. (Perrino et al., 2024) • The family was educated on supporting communication skills. (Perrino et al., 2024) • Education was provided. (Perrino et al., 2024)	An increase was observed in the child's communication attempts and responses.

Conclusion

Costello syndrome is a rare childhood condition that attracts clinical attention due to its multisystem involvement as a RASopathy. Caused by mutations in the HRAS gene, this syndrome presents a broad clinical spectrum ranging from cardiac complications to neurodevelopmental issues and an increased risk of malignancy. As demonstrated in the case report, early-onset feeding difficulties, developmental delays, and characteristic phenotypic features provide important clues for diagnosis. However, definitive diagnosis is established through genetic analysis, and a multidisciplinary approach significantly impacts patients' quality of life.

Recognizing Costello syndrome in childhood is critically important for the early detection of cardiac and oncologic complications as well as for the timely initiation of developmental support programs. This case report emphasizes the clinical features of this rare syndrome and highlights the necessity of a multidisciplinary approach in its management. Nursing care plays a crucial role in addressing feeding difficulties, providing developmental support, and educating families throughout the disease course. In conclusion, the follow-up of children with Costello syndrome requires comprehensive evaluation including genetic, pediatric, cardiologic, and oncologic assessments, alongside nursing care as an integral component of holistic management. It is recommended that studies focusing on nursing care be expanded, and evidence-based example care plans within a scientific and theoretical framework be increased in the literature.

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Exploring Burnout Prevention in Hungarian Pharmacy Education: An Exploratory Curriculum Mapping Study

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ABSTRACT

Burnout among healthcare professionals has received increasing attention due to its impact on professional wellbeing, patient safety, and workforce sustainability. Although pharmacists play a key role in healthcare systems, research on burnout prevention has mainly focused on practicing professionals. Emerging evidence suggests that preventive approaches should begin earlier, during professional education. Pharmacy programs, characterized by intensive workload and high professional expectations, provide a context for examining how wellbeing and burnout prevention are addressed within the curriculum. An exploratory curriculum mapping approach was used to examine how burnout prevention and related wellbeing competencies appear in Hungarian pharmacy education. Publicly available curriculum documents and course descriptions were reviewed across the four universities offering pharmacy programs: Budapest, Debrecen, Szeged, and Pécs. The analysis searched for explicit and implicit references to burnout prevention, resilience, stress management, professional identity formation, and broader wellbeing competencies. Explicit burnout-prevention content was largely absent from the formal curriculum. Across institutions, at most one elective course addressed resilience or stress management, while no mandatory course defined burnout prevention as a distinct learning objective. Approximately 10–15% of elective courses included indirectly relevant topics such as communication, ethics, or patient counselling. In contrast, more than 85% of the core curriculum focused on pharmaceutical sciences, clinical knowledge, and practice management. Wellbeing-related competencies appear in Hungarian pharmacy curricula primarily in implicit and fragmented ways. Pharmacy education therefore represents an underutilized opportunity for early burnout prevention. Curriculum mapping may support the future integration of structured wellbeing competencies into pharmacy education.

Keywords – Burnout prevention, Pharmacy education, Curriculum mapping, Student wellbeing, Health professions education

I. INTRODUCTION

Across health professions worldwide, increasing attention is being directed toward the phenomenon of burnout. Burnout affects not only individual professional wellbeing but is also closely linked to patient safety, quality of care, professional performance, and the long-term sustainability of the healthcare workforce.

Although pharmacists play a crucial role within healthcare systems, most research on burnout prevention has focused primarily on practicing professionals. In this presentation, I argue that prevention should begin much

earlier—during professional education itself. To explore this perspective, I present findings from an exploratory curriculum analysis conducted within the context of Hungarian pharmacy education.

Burnout Among Pharmacists: Pharmacists work in environments characterized by high cognitive workload, time pressure, significant legal and ethical responsibility, and intensive patient communication. These factors alone may substantially increase the risk of burnout.

International research also suggests that symptoms of burnout may emerge already during professional training, particularly in demanding health professions programs with extensive academic workload. This raises an important question: to what extent does university education prepare pharmacy students not only professionally, but also mentally and emotionally for the challenges of their future careers?

Education as a Preventive Opportunity: Traditionally, health professions education has focused primarily on the transmission of knowledge and professional competencies. However, there is growing recognition that education itself can serve as a preventive space.

In many programs, learning is often structured around the completion of courses and the achievement of grades. Yet professional development also involves the acquisition of less visible competencies such as self-reflection, coping strategies, and emotional resilience.

Pharmacy education may offer a particularly valuable context for integrating these elements. As students gradually develop their professional identity, they may also benefit from learning skills related to stress management, reflective practice, and resilience. However, before such integration can occur effectively, it is necessary to understand how and where these topics currently appear within existing curricula [1].

Problem Statement and Research Gap: In many educational contexts, wellbeing and burnout prevention are not explicitly articulated as curricular objectives. Instead, they tend to appear indirectly within certain courses—often those focusing on communication, ethics, or professional interaction [2, 3].

While these elements may contribute to student wellbeing, their fragmented presence limits their potential preventive impact. Moreover, it becomes difficult to assess whether students are truly being prepared to manage the psychological and professional demands of their future careers [4].

These considerations provided the motivation for conducting a systematic curriculum mapping exercise.

II. Research Aim

The aim of this study was to explore how themes related to burnout prevention, resilience, stress management, professional identity formation,

and general wellbeing competencies are represented within Hungarian pharmacy curricula.

The purpose of the analysis was not to evaluate or rank institutions, but rather to provide an overview of the current educational landscape and to identify broader patterns within the system.

III. Materials and Method

A. Methodology: Curriculum Mapping

An exploratory curriculum mapping approach was employed. Publicly available curriculum documents and course descriptions were reviewed across the four Hungarian universities offering pharmacy education.

The analysis focused on identifying both explicit and implicit references to the following themes:

– burnout prevention,
– resilience,
– stress management,
– professional identity formation,
– general wellbeing-related competencies.

The approach was qualitative and descriptive in nature, aiming to identify patterns, gaps, and areas of potential development within the curricula.

B. Institutions Included in the Analysis

The analysis covered the four Hungarian universities offering pharmacy education:

– Faculty of Pharmacy, Semmelweis University, Budapest [5],
– Faculty of Pharmacy, University of Debrecen, Debrecen [6],
– Faculty of Pharmacy, University of Pécs, Pécs [7, 8],
– Faculty of Pharmacy, University of Szeged, Szeged [9].

All four institutions offer integrated, pre-licensure pharmacy programs operating within similar regulatory and structural frameworks. This comparability allowed for a meaningful exploratory overview of curricular patterns across institutions.

IV. Results

Explicit Content

The analysis suggests that explicit burnout-prevention content is largely absent from current curricula.

Across institutions, at most one elective course directly refers to topics such as resilience or stress management. Within the mandatory core curriculum, no course was identified that explicitly defines burnout prevention as an independent learning objective.

Implicit Content

At the same time, several indirectly relevant elements were identified.

Approximately 10–15 percent of elective courses include themes related to communication, ethics, or patient counselling. These courses may indirectly support student wellbeing and professional development.

However, their presence is not consistent across institutions, and they are not embedded within a clearly defined wellbeing or prevention framework.

Focus of the Core Curriculum

More than 85 percent of the mandatory curriculum focuses on pharmaceutical sciences, clinical knowledge, and practice management.

This emphasis is professionally justified and reflects the strong scientific foundation of pharmacy education. Nevertheless, the findings also illustrate that mental wellbeing and burnout prevention are currently not prominent priorities within the formal curriculum structure.

V. Discussion

The findings suggest that wellbeing-related competencies are present within pharmacy curricula, but mostly in implicit, fragmented, and institution-specific ways.

Pharmacy education therefore represents a critical—yet currently underutilized—opportunity for early burnout prevention.

Importantly, these observations should not be interpreted as shortcomings of individual institutions. Rather, they point toward a broader structural and systemic issue within health professions education.

VI. Conclusions

Curriculum mapping can serve as a valuable tool for making wellbeing visible within health professions education.

Looking forward, there may be a need to integrate structured and longitudinal wellbeing competencies into pharmacy education, ideally as a natural component of professional identity formation.

We hope that this exploratory analysis can contribute to a broader professional dialogue about how pharmacy education can evolve toward a more sustainable and student-centered model of training.

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