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Thyroid Hormone Resistance Syndromes

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Statement of the Problem: Thyroid hormone resistance (THR) syndromes are rare but often misdiagnosed disorders affecting metabolic homeostasis. Understanding their genetic basis is crucial for effective treatment.

Methodology & Theoretical Orientation: This study analyzed 50 THR patients using whole-exome sequencing and functional assays to identify mutations in the thyroid hormone receptor beta (THRB) gene.

Findings: Novel THRB mutations were identified in 30% of patients. Functional assays demonstrated reduced receptor binding affinity and impaired transcriptional activity.

Conclusion & Significance: Genetic testing should be integrated into clinical protocols for THR diagnosis. These findings may guide the development of targeted therapies

Biography

Dr. Priya Sharma is a geneticist and endocrinologist with extensive expertise in rare thyroid disorders. Her groundbreaking research has identified novel mutations and their implications for disease mechanisms. Dr. Sharma is dedicated to improving diagnostic tools and personalized treatment for thyroid hormone resistance syndromes.

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