

## DGCR8 Microprocessor Subunit Mutation and Expression Deregulation in Thyroid Lesions

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*Deregulation of microRNA (miRNA) processing is a driver event in several tumours including thyroid cancer. DiGeorge Critical Region 8 (DGCR8) gene holds a critical role in miRNA biogenesis, as a microprocessor complex component, and in the development of the thyroid. Previous studies identified a DGCR8 mutation – the variant c.1552G>A p.(E518K) – in cases of thyroid cancer and proposed to cause a familial*

*syndrome characterized by multinodular goitre (MNG) and schwannomatosis. The goal of this study was to characterize the variant p.(E518K) of DGCR8 in thyroid lesions and evaluate its expression.*

*A series of thyroid lesions were evaluated by sequencing for the c.1552G>A p.(E518K) variant. When frozen tissue was available, DGCR8 mRNA expression was analysed by qPCR. Formalin-fixed paraffin-embedded tissues were studied for DGCR8 immunoeexpression.*

*We present for the first time the p.(E518K) mutation in a case of poorly differentiated thyroid carcinoma and present the deregulation of DGCR8 expression at mRNA level in follicular-patterned tumours. The obtained data solidify DGCR8 as another important player of miRNA-related gene mutations in thyroid tumorigenesis, particularly in follicular-patterned thyroid tumours.*

**Keywords:** miRNA; thyroid cancer; microprocessor complex; DGCR8; p.(E518K).