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Non-invasive lung cancer diagnosis by detection of GATA6 and NKX2-1 isoforms in exhaled breath condensate

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Abstract

© 2016 The Authors. Published under the terms of the CC BY 4.0 license Lung cancer (LC) is the leading cause of cancer-related deaths worldwide. Early LC diagnosis is crucial to reduce the high case fatality rate of this disease. In this case-control study, we developed an accurate LC diagnosis test using retrospectively collected formalin-fixed paraffin-embedded (FFPE) human lung tissues and prospectively collected exhaled breath condensates (EBCs). Following international guidelines for diagnostic methods with clinical application, reproducible standard operating procedures (SOP) were established for every step comprising our LC diagnosis method. We analyzed the expression of distinct mRNAs expressed from GATA6 and NKX2-1, key regulators of lung development. The Em/Ad expression ratios of GATA6 and NKX2-1 detected in EBCs were combined using linear kernel support vector machines (SVM) into the LC score, which can be used for LC detection. LC score-based diagnosis achieved a high performance in an independent validation cohort. We propose our method as a non-invasive, accurate, and low-price option to complement the success of computed tomography imaging (CT) and chest X-ray (CXR) for LC diagnosis.

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Keywords

EBC, GATA6, lung cancer, molecular diagnostics, NKX2-1