

# The role of L1 retrotransposon in circulating tumor cells

## Die Rolle von L1 Retrotransposon in zirkulierenden Tumorzellen

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**Introduction:** L1 belongs to the family of non-long terminal repeat retrotransposons. It comprises about 17% of the human genome and consists of two different open reading frames (ORFs). The ORF1 has nucleic acid chaperone activity while the ORF2 contains both endonuclease and reverse transcriptase activities. Retrotransposons can induce mutations or regulate the expression of specific genes. The present study aimed to analyze the epigenetic profile of L1 between circulating tumor cells (CTCs) and non-cancerous samples.

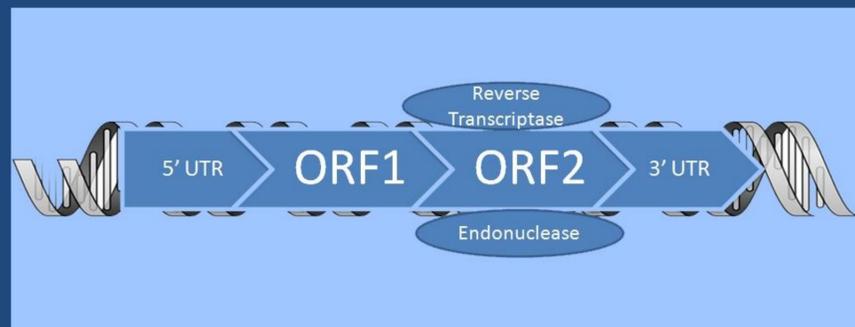


Figure 1: L1 DNA Structure

**Conclusion:** The L1 retrotransposon is found in all mammals and is the only active element in the human genome. According to literature and experimental data, the ORF2 is involved in stemness transcription factors. The present study demonstrated that ORF2 RNA expression is higher in CTCs than in normal cells, indicating once again the role of L1 in carcinogenesis.

**Materials & Methods:** Blood samples were collected from healthy donors and patients, suffered from prostate, lung, breast, colon and pancreatic cancer. Pancytokeratin positive selection was performed for cancer samples, while CD45 negative selection was followed for healthy individuals. DNA and RNA was extracted from the above samples and qPCR was performed for L1-ORF1 and L1-ORF2. Data were analyzed by using DeltaCt method and 18S rRNA as housekeeping gene. One-way ANOVA was performed to compare the normal and cancer samples.

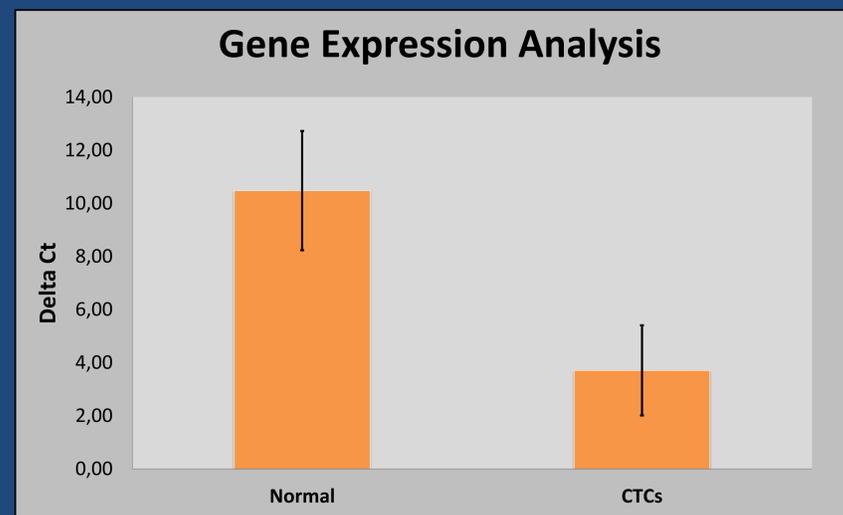


Figure 2: Comparative qPCR analysis of L1-ORF2 expression in normal and different cancer samples. A lower DeltaCt indicates higher gene expression.

**Results:** The DNA analysis revealed the presence of both L1-ORF1 and L1-ORF2 in all samples. The gene expression analysis demonstrated that L1-ORF1 is not expressed in the majority of the samples, either normal or cancerous. Regarding the L1-ORF2, it was expressed in all samples, however, it was overexpressed in CTCs ( $p=0.00089 < 0.5$ ).

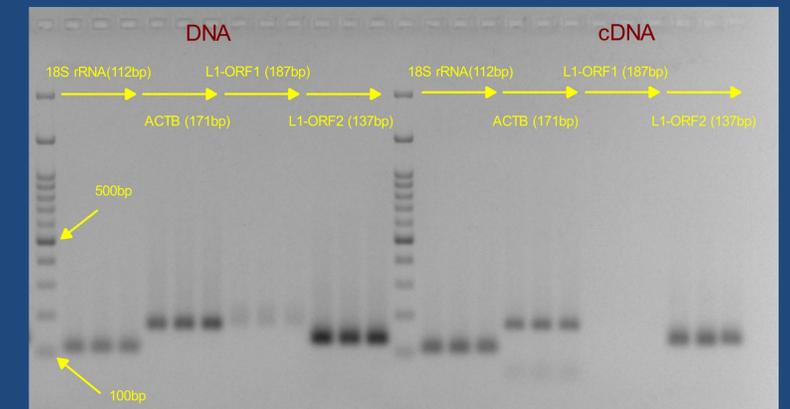


Figure 3: Electrophoresis of PCR products of a lung cancer patient

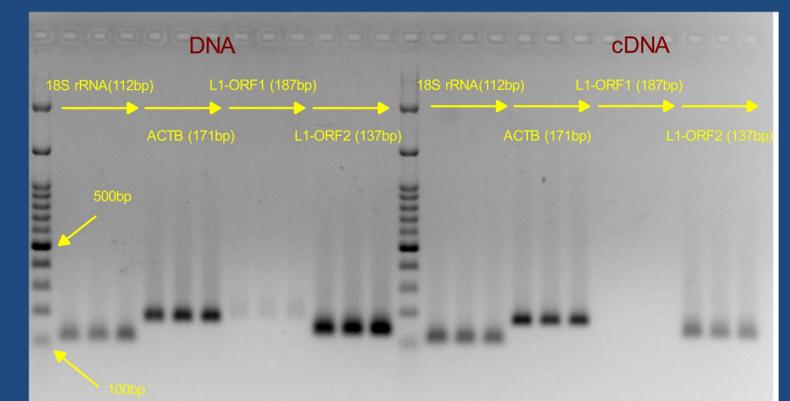


Figure 4: Electrophoresis of PCR products of a healthy individual

### Selected References:

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### Disclosure of Potential Conflicts of Interest

None of the authors of the above study has declared any conflict of interest