高通量基因测序技术检测外周血循环肿瘤DNA基因突变在非小细胞肺癌中的应用

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Title: **Application of high-throughput sequencing technology in the detection of peripheral blood circulating tumor DNA mutation for non-small cell lung cancer patients**

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关键词: 非小细胞肺癌; 外周血肿瘤DNA; 高通量基因测序

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摘要: 目的：探究高通量基因测序技术检测非小细胞肺癌外周血循环肿瘤DNA基因突变的应用价值。方法：临床纳入2017年1月至2018年9月在我院就诊的40例晚期非小细胞肺癌患者作为研究对象，所有患者入院后均经肺组织活检或气管镜检查确诊为晚期非小细胞肺癌。对患者进行病理组织石蜡切片DNA（tDNA）检测，并采集患者肘静脉血使用高通量基因测序技术检测患者外周血循环肿瘤ctDNA基因情况。对比分析tDNA与ctDNA检测对患者DNA基因突变的准确性，探讨非小细胞肺癌患者进行高通量基因测序技术检测外周血循环肿瘤DNA基因突变的应用价值。结果：40例非小细胞肺癌的外周血循环肿瘤DNA基因突变检测与组织石蜡切片比较，两种方法检测率差异无统计学意义（P>0.05）。在高通量基因测序技术检查外周血循环肿瘤DNA中，21外显子测序结果：61号替代突变2573G→T，62/63/68号替代突变L858R（2573T→G）。19外显子测序结果：50号样品突变为del E746→A750+2235G→A，60号样品突变为del E746→A750，70号样品突变为del L747→T751，80号样品突变为del L747→S752＋2257C→T。结论：非小细胞肺癌外周血循环肿瘤DNA基因突变进行高通量基因测序技术对具体的基因突变或缺失具有较高准确性，可实时监测肿瘤DNA基因突变情况，且具有无创性、可重复应用等优点。

Abstract: Objective：To explore the application value of high-throughput sequencing technology in the detection of peripheral blood circulating tumor DNA mutation for non-small cell lung cancer (NSCLC) patients.Methods：40 advanced NSCLC patients confirmed by the tissue biopsy from January 2017 to September 2018 in our hospital were selected.Upon admission to hospital，the tDNA (transferred DNA) mutations were detected by tissue paraffin section while the ctDNA (circulating tumor DNA) mutations were detected by high-throughput sequencing technology.The diagnosis accuracy based on ctDNA and tDNA was compared to discuss the application value of high-throughput sequencing technology in the peripheral blood circulating tumor DNA mutation.Results：For 40 NSCLC patients，the detection rate of ctDNA mutations was not significantly different from that of tDNA in the tissue paraffin section (P>0.05).Through the high-throughput sequencing technology，the exon21 indicated including type 61/62/63/68.The EGFR exon 19 test indicated that type 50 gene mutation was del E746→A750+2235G→A，type 60 gene mutation as del E746→A750，type 70 gene mutation as del L747→T751 and type 80 gene mutation as del L747→S752＋2257C→T.Conclusion：For NSCLC patients，the high-throughput sequencing technology has the higher detection accuracy in the gene mutations or deficiency，which it is featured as timeliness，non-invasive surgery and repeated application.

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