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應用 HRM 分析方法快速偵測急性骨髓性白血病患 IDH1 與 IDH2 基因突變

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Application of the rapid HRM analysis to detected IDH1 and IDH2 mutation in acute myeloid leukemia

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BACKGROUND:
Isocitrate dehydrogenase 1 (IDH1) and IDH2 mutations and polymorphism are reported in 5% to 15% of acute myeloid leukemia (AML) cases, with R132 of IDH1 and R172 of IDH2 known to be clinically significant. High resolution melting (HRM) is a novel screening method that enables rapid identification of mutation positive DNA samples. In this study, we use rapid HRM analysis for the detection of IDH1 and IDH2 mutation in AML.
METHODS and MATERIALS:
We successful establish of IDH1 and IDH2 mutation and polymorphism by HRM analysis. 11 patients diagnosed with AML were examined. The research project was reviewed and approved by the IRB committee of our hospital (DMR99-IRB-108). All mutations are confirmed with directly sequencing.
RESULTS:
Herein, we established a rapid detection molecular diagnosis about IDH1 and IDH2 mutation assays for HRM. Analysis of a series of 11 patient specimens revealed 2 positive for IDH1 mutation and 4 positive for IDH2 mutation.
CONCLUSIONS:
This study demonstrates that the HRM assay could rapid, convenient, and versatile assays for screening and confirmation of alterations in IDH1 and IDH2. We expect to set up the molecular diagnostic technique for clinical laboratory service.

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利用 HRM 分析方法快速偵測早發型巴金森氏病風險基因檢測

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Rapid detection of early-onset Parkinson's Disease risk gene by high-resolution melting curve analysis

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BACKGROUND:
Parkinson's disease (PD) is the second most frequent neurodegenerative disorder after Alzheimer's disease. Approximately 5~10% of patients with the clinical picture of PD carry a mutation in one of the known genes that cause monogenic forms of the disorder. High-resolution melt (HRM) curve analysis is a rapid and non-gel-based method. This study was to assess the early-onset Parkinson's Disease(EOPD) risk gene mutation of the HRM analysis.
METHODS and MATERIALS:
In the present study was aimed to screen the clinical diagnosed familial EOPD for *Parkin*, *PINK1*, and *ATP13A2* detected by HRM method. Screening 34 patients with clinical diagnosis of Parkinson's disease (also has a family history and early-onset). All mutations were confirmed with PCR-Sequence.
RESULTS:
There were seven hotspot finding by HRM distributed on *Parkin* (Ser167Asn, Arg366Trp, and Val380Leu)、*PINK1* (Pro209Ala, Ala340Thr, and Met341Val), and *ATP13A2*(Ala746Thr). In additional, we found two of patients with EOPD has new mutation *PINK1* (Val350Leu heterozygote) and *ATP13A2* (Asn739Asn heterozygote).
CONCLUSIONS:
HRM assay offers the advantage of high throughput, single step, cost effectiveness and can be utilized as a rapid screening method for detection of *Parkin* (Ser167Asn, Arg366Trp, and Val380Leu), *PINK1* (Pro209Ala, Ala340Thr, and Met341Val), and *ATP13A2* (Ala746Thr) mutations in a clinical setting. Finally, the clinical association of *PINK1* (Val350Leu) and *ATP13A2* (Asn739Asn) with EPOD should need more clinical data to be demonstrated.