Influence of \textit{ERCC2} gene polymorphisms on the treatment outcome of osteosarcoma


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\textbf{ABSTRACT}. We conducted a prospective study to investigate the role of \textit{ERCC2} gene polymorphisms on the outcome of cisplatin-based treatment in patients with osteosarcoma. A total of 115 patients with osteosarcoma were included in our study. Genotyping of \textit{ERCC2} Asn312Asp (rs1799793) and Lys751Gln (rs13181) was performed using a matrix-assisted laser desorption/ionization time-of-flight mass spectrometry method. Of the 115 patients, 78 showed complete or partial response to chemotherapy, with a response rate of 67.85\%. Our study suggested that the AA genotype of \textit{ERCC2} Asn312Asp was associated with a better response to chemotherapy, and the related adjusted OR (95\%CI) was 4.85 (1.06-42.71). By Cox proportional hazards model analysis, we found that the AA genotype of \textit{ERCC2} Asn312Asp was associated with longer overall survival of patients with osteosarcoma when compared with the GG genotype, and the hazards ratio (95\%CI) for the AA genotype was 0.65 (0.27-1.47). In conclusion, our study found that the \textit{ERCC2} Asn312Asp gene polymorphism likely plays an important role in influencing the chemotherapy response and overall survival of patients with osteosarcoma.

\textbf{Key words}: \textit{ERCC2}; Polymorphism; Osteosarcoma; Treatment outcome