(p＝0.02) と *MUTYH* 低活性型 (p＝0.0001) の遺伝子型が MM の生存期間に負の影響を与える因子として抽出された. **【結　論】** BER 遺伝子の低活性群では, 塩基除去修復能が低下し, MM の発症に関与することが示唆された. また, これらの遺伝子多型は MM の臨床像と関連し, MM の進展や予後にも影響を与えることが示唆された.

34. The association of polymorphism in DNA BER genes OGG1, XRCC1, APE1 and MUTYH with the risk/clinical characteristics of myelodysplastic syndromes (MDS) in Japanese patients

Batchimeg Norjmaa,[1] Takayuki Saitoh,[1] Atsushi Iwasaki,[1] Yasuhiro Nitta,[1] Hiroshi Handa [2] and Hirokazu Murakami[1]

（1　Department of Laboratory Sciences, Gunma University Graduate School of Health Sciences）

（2　Department of Hematology, Gunma University Hospital）

【Background】 Base excision repair (BER) is mainly responsible for the correction of small base changes of DNA damage. BER pathway involved many enzymes including OGG1, XRCC1, APE1 and MUTYH. The single nucleotide polymorphisms of their genes have been reported to relate with several cancers. The defective DNA repair is associated with an increased risk of various cancers including hematologic malignancies. However, it is unclear these polymorphisms alter the susceptibility and clinical outcome of MDS patients. The aim of this study is to evaluate the association of polymorphisms in gene encoding four key proteins of DNA base excision repair (BER): *OGG1*Ser326Cys, *XRCC1*Arg399Gln, *APE1*Asp148Glu and *MUTYH* Gln324His with the susceptibility and clinical features of MDS. **【Methods】** Our study included 106 MDS patients［median 65.3 years, range 17.0–86.5 years; male/female 71/35; RCUD (n＝25), RARS (n＝7), RCMD (n＝19), RAEB-1 (n＝12), RAEB-2 (n＝12), MDS-U (n＝9), and others (n＝22)］and 192-health control group. Genetic polymorphisms in BER pathway genes were examined using PCR and polymerase chain reaction-restriction fragment length polymorphism (PCR-RFLP) technique. **【Results】** There was no significant difference in the allele and genotype frequencies of BER gene polymorphisms between the MDS patients and the control group. In the analysis of clinical characteristics, *XRCC1* Arg/Arg genotype was significantly associated with lower Hb level (8.78±2.41 g/dL vs. 10.0±2.08 g/dL, p＜0.05) and higher frequency of transformation to leukemia (21.6% vs. 7%, p＜0.05). *XRCC1* non-Arg/Arg genotype was associated with the secondary MDS (33.3% vs 12.7%, p＜0.05). In addition, the frequency of blood transfusion was significantly associated with *OGG1* non-Ser/Ser genotype (63.6% vs 20.7%, p＜0.01). In contrast, the polymorphisms in *APE1* Asp148Glu and *MUTYH* Gln324His did not have statistically significant differences in the clinical features of MDS. **【Conclusion】** Our findings suggest that *OGG1* and *XRCC1* gene polymorphisms may be associated with the clinical features of MDS in Japanese patients.