

类核沉降法对三个着色性干皮病(XP)家系成员DNA修复能力的测定及杂合子的检出*

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摘要 应用类核沉降法, 分析了三个着色性干皮病家系中35名成员外周血淋巴细胞在紫外线(2.5μJ/mm²)照射和MNNG(2μg/ml)损伤后DNA修复能力。结果表明, 6名XP患者和9名杂合子DNA损伤手20小时尚不能完成修复, 他们11小时DNA修复率均值分别为UV, 0.61±0.13, 0.59±0.15和MNNG, 0.44±0.15, 0.46±0.16, 与家系中10名非血缘亲属正常人DNA修复率均值为UV, 0.96±0.07, MNNG, 0.71±0.07, 相比, 差异非常显著(P<0.01), 而XP患者和杂合子DNA修复率则无显著性差异(r>0.05)。提示, 类核沉降技术可能为XP家系中杂合子成员检出提供一种有希望的方法。

关键词 [类核沉降法](#), [DNA损伤修秒](#), [着色性干皮病](#), [杂合子](#)

分类号

DNA Repair Capacity Measurement and Possibility of Heterozygote Detection Using

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Abstract

The nucleoid sedimentation test was used to analyse the DNA repair capacity in 35 members taken from 3 xeroderma pigmentosum (XP) families after their peripheral blood lymphocytes being exposed to UV irradiation or N-methyl-N-nitrosoguanidine (MNNG) at a dose of 2.5 μJ/mm² and 2 μg/ml respectively. The results showed that the 6 XP patients all failed to repair their damaged DNA even after 20-hour incubation at 37°C. The repair ratios were 0.61±0.13 for UV and 0.44±0.15 for MNNG while those of non-related normal members usually reached 0.96±0.07 and 0.71±0.07 for both UV and MNNG after 11-hour incubation. It is interesting to note that 9 obligatory heterozygotes (parents and grandparents of XP patients) and 4 highly possible heterozygotes (blood relatives of XP patients) also showed very poor DNA repair capacity: they could not accomplish their repair process even after 20-hour incubation. It has been showed that the nucleoid sedimentation test can probably be used to pick out the heterozygotes in XP families.

Key words [Nucleoid sedimentation](#) [DNA damage repair](#) [Xeroderma pigmentosum](#) [Heterozygotes](#)

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