

# 人类核仁形成区结构性变异与随体联合的研究<sup>1)</sup>

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**摘要** 本文以硝酸银染色法研究了200对正常夫妇及200对唐氏综合征(DS)患者双亲淋巴细胞染色体核仁形成区(NOR)。结果发现双核仁形成区(dNOR)的检出率在两组人群中均为0.5%；随体联合(SA)的结果表明，负有dNOR染色体的SA频率并不随其核糖体RNA(rRNA)基因含量的明显增加而上升，提示除rRNA基因含量及其活性外，尚有其他因素影响SA的形成。dNOR携带者与DS的发生亦无明显关系。

**关键词** [双核仁形成区,随体联合](#)

分类号

## Study on Structural Variation of Nucleolar Organizer Regions (NOR) and the Satellite Association (SA)

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### Abstract

To further illustrate the biological significance of variant NOR (including double NOR-dNOR and triple NOR-tNOR) and satellite association (SA) and the relationship to meiotic non-disjunction (NDJ), this study was carried out in the normal population and the parents of patients with Down Syndrome (DS). Of 800 individuals, 4 dNOR carriers, two of them from the control and two from the non-informative experiments were detected. SA data for 3 out of the 4 dNOR carriers showed no statistically significant increase in SA when dNOR was present in the cell even in case 4 where two dNOR were found in one cell. However, case 3 demonstrated significant decrease in SA when dNOR was present, although it showed that the overall incidence of dNOR was higher in this carrier than that in the other three. It is concluded that dNOR can be detected on any acrocentric chromosome and not be related to NDJ of chromosome, which is a heritable variant that does not affect phenotype and that there are two kinds of dNOR variants, one being from the translocation of the short arm of acrocentric chromosome, the other from non-translocation.

**Key words** [double Nucleolar Organizer regions \(dNOR\)](#) [Satellite association \(SDA\)](#)

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