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综述

NPM1基因突变与急性髓细胞白血病

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摘要

NPM1(nucleophosmin,又称B23, numatrin或N038)是一种主要定位于核仁，可在核仁与胞浆之间穿梭的核磷蛋白。第12外显子突变导致NPM1胞浆异位从而发生肿瘤转化。NPM1突变与正常核型的急性髓细胞白血病、成年女性、多系受累、CD34-、 FLT3-ITD、独特的临床表现及良好的预后有关。对于NPM1+/FLT3-ITD-患者，移植与否对预后无差别。NPM1突变导致白血病发生的机制尚不清楚。

关键词 核磷蛋白；急性髓细胞白血病；基因突变

分类号

NPM1 gene mutation and acute myeloid leukemia

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Abstract

NPM1 (nucleophosmin, also named as B23, numatrin or N038) is a nucleophosmin that is localized mainly in the nucleolus, continuously shuttles between the nucleus and cytoplasm .Mutations in exon 12 cause cytoplasmic NPM1 localization, and consequently contribute to tumour development.NPM1 mutations correlate with normal karyotype acute myeloid leukemia, adult female, multilineage involvement, CD34 negativity,FLT3-ITD,special clinical feature and better prognosis.For the patients with NPM1-mutated/FLT3 ITD-negative, there is no different prognosis between allogeneic SCT and non-allogeneic SCT. Now how mutated NPM1 contributes to leukemogenesis still remains to be explored.

Key words NPM1（nucleophosmin） acute myeloid leukemia (AML) gene mutation

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