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The spectrum of α and β -thalassemia mutations of the Li people in Hainan province of China

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This study examines the frequency and spectrum of α and β -thalassemia (thal) mutations of the Li people in Hainan Province of China. We have analyzed by genotyping a sample of 8600 subjects of the Li people and found that 53.45% subjects have only α -thal mutations with high frequencies of α 4.2 and α 3.7 but fewer SEA mutation; 3.83% have β -thal mutations all identified to be 41/42 (TCTT); whereas 7.99% carry both α -thal and β -thal mutations. We also examined 9800 subjects of the Han people and the result showed 12.16% subjects have only α -thal mutations with SEA and α 3.7 the most frequent mutation types, 6.11% have only β -thal mutations of 7 types, whereas 4.85% carry both α -thal and β -thal mutations. Our study demonstrated that the Li people in Hainan province have a high incidence of α 4.2 and α 3.7 thalassemia, low frequencies of α -thal SEA and a novel β -mutation, 41/42 (TCTT). We provide the complete spectrum of α -thal and β -thal mutations and a strategy for accurate molecular diagnostic testing in the Li people in Hainan province of Southern China.

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Safety of intrathecal chemotherapy in the management of hematological malignancies

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Treatment of leptomeningeal metastasis (LMD) remains challenging due to advanced systemic disease at presentation and limited treatment options. Intrathecal chemotherapy is one of the options actually used. Besides the hematological malignancies, the prophylactic use of this procedure is common in some entities. Unless, there is no complete knowledge of the toxicity and security of this practice, different factors could influence in the presence or not of adverse events. These factors could be the way of administration (omaya reservoir or lumbar puncture), the presence or not of disease (prophylaxis or treatment), the drug we use (triple therapy or liposomal cytarabine or metotrexate), the type of malignancy (lymphoma, leukemia or myeloma). It is necessary to know all these features with the aim of checking the security of a procedure that nowadays is a common alternative of treatment of a serious complication of hematological malignancies.

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