

论著

我国辽宁地区遗传性粘多糖贮积症 I 型患者 α -L-艾杜糖醛酸酶基因突变的研究

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摘要 目的: 探讨我国粘多糖贮积症I型患者 α -L-艾杜糖醛酸酶基因的突变情况。方法: 采用PCR-SSCP和DNA测序的方法检测我国辽宁地区10个粘多糖贮积症I型家系 α -L-艾杜糖醛酸酶基因的突变类型。结果: ①发现我国辽宁地区粘多糖贮积症I型患者 α -L-艾杜糖醛酸酶基因存在2种新的突变类型R363H和880+g-c。②同时发现3种多态性位点R105Q、L118和A361T。结论: 我国辽宁地区粘多糖贮积症I型患者 α -L-艾杜糖醛酸酶基因的突变情况不同于其他国家和地区, 但其多态性与Scott等报道的欧裔患者情况相似 [1]。

关键词 [粘多糖累积病 I 型](#); [艾杜糖醛酸酶](#); [突变](#); [多态现象](#)

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The mutation of α -L-iduronidase gene for mucopolysaccharidosis type I in Liaoning district populations

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Abstract

AIM: To investigate the mutataion type and polymorphism site in the α -L-iduronidase (IDUA) gene of Liaoning district MPS-I patients. METHODS: The mutation type and polymorphism site in the IDUA gene of Liaoning district mucopolysaccharidosis type I (MPS-I) patients were detected by PCR-SSCP and DNA sequencing. RESULTS: ① 2 new mutations: R363H, 880+g-c in the IDUA gene of Liaoning district MPS-I patients were found. ② There were 3 polymorphism sites: R105Q, L118 and A361T in the IDUA gene of Liaoning district MPS-I patients. CONCLUSIONS: The mutation type in the IDUA gene of Liaoning district MPS-I patients is different from that of other countries and districts, while the polymorphism site in the IDUA gene of Liaoning district MPS-I patients is the same as that of other countries.

Key words [Mucopolysaccharidosis I](#) [Iduronidase](#) [Mutation](#) [Polymorphism](#)

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