

GC-MS方法诊断新生儿苯丙酮尿症

邓春晖,胡耀铭,胡克季,张雅芬,顾学范

复旦大学分析测试中心.上海(200433);上海新华医院

收稿日期 修回日期 网络版发布日期 接受日期

摘要 用甲醇提取血样中的苯丙氨酸(Phe)、酪氨酸(Tyr), 提取液离心去蛋白质, 用正丁醇将其中的苯丙氨酸和酪氨酸丁酯比, 再用三氟醋酸酐酰化后进行GC-MS分析, 测定Phe, Tyr特征离子峰面积, 用外标法计算出Phe和Tyr的摩尔浓度, 根据Phe和Tyr摩尔浓度比值来诊断新生儿苯丙酮尿症(PKU)。

关键词 [色谱质谱法](#) [苯丙酮尿症](#) [氨基酸](#) [苯丙氨酸](#) [酪氨酸](#)

分类号 [0658](#)

GC-MS screening of phenylketonuria in newborns

Deng Chunhui, Hu Yaoming, Hu Keji, Zhang Yafen, Gu Xuefan

Fudan Univ, Ctr Anal & Measurement, Shanghai(200433)

Abstract Phenylalanine(Phe) and Tyrosine(Tyr) were extracted from dry blood spots with 0.1% mol/L HCl-methanol. The proteins in the extraction were removed by centrifuging. Phe and Tyr were treated with 3.0 mol/L HCl-butanol. Phe and tyrosine butyl esters were treated with trifluoroacetic anhydride. The derivatives from Phe and Tyr were analyzed by GC-MS. The contents of the two amino acids in blood samples were determined by calculating the areas of their derivatives with reference to an exterior standard. Finally the diagnosis of PKU was made on the basis of the molar content ratios of Phe to Tyr.

Key words [CHROMATOGRAPHY-MASS SPECTROGRAPHY](#) [AMINOACIDURIA](#) [AMINO ACID](#) [PHENYLALANINE](#) [TYROSINE](#)

DOI:

通讯作者

扩展功能

本文信息

▶ [Supporting info](#)

▶ [PDF\(0KB\)](#)

▶ [\[HTML全文\]\(0KB\)](#)

▶ [参考文献](#)

服务与反馈

▶ [把本文推荐给朋友](#)

▶ [加入我的书架](#)

▶ [加入引用管理器](#)

▶ [复制索引](#)

▶ [Email Alert](#)

▶ [文章反馈](#)

▶ [浏览反馈信息](#)

相关信息

▶ [本刊中包含“色谱质谱法”的相关文章](#)

▶ 本文作者相关文章

- [邓春晖](#)
- [胡耀铭](#)
- [胡克季](#)
- [张雅芬](#)
- [顾学范](#)