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• 医学新闻 •

北京协和医院儿科成功绘制我国溶酶体疾病谱

北京协和医院儿科历经 14 年对 1226 例溶酶体病患者进行临床研究和总结, 完成了国内首次单中心大样本病例总结和国际罕见单人种大样本分析, 首次报道了中国人溶酶体病的疾病谱。溶酶体病是由于溶酶体内 60 余种酸性水解酶中的任何酶发生缺陷所造成的疾病, 目前已知的病种超过 50 余种, 其中绝大多数属于常染色体隐性遗传病。溶酶体病十分罕见, 总发生率不详。本项科研成果是国内最大样本的总结, 为我国溶酶体病的发生率统计提供了关键数据。在该项目的促进下, 北京协和医院溶

酶体病诊断技术不断提高, 实现了从单纯酶活性测定到酶活性测定结合基因突变家系分析诊断技术的突破, 更加有效地保证了诊断的准确率。该项目组还参与了全球多中心的临床药理 III 期研究, 参与制定国内仅有的 2 个溶酶体病的专家共识, 帮助其他两个诊断中心培训数名诊断试验技术人员。北京协和医院儿科以高质量的诊断和产前诊断水平, 得到国内业界的高度评价。

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