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吴柏林教授的主要研究方向遗传学和基因组医学，基因诊断和分子病理。最新的研究包括率先研发了可用于临床的全基因组高分辨高通量微阵列基因芯片，成功地发现了两个与孤独症发病相关的可重复发生的遗传病因，在与基因组失衡相关的出生缺陷和发育性疾病的遗传病因鉴定及研发相应的临床基因诊断的转化医学研究方面走在世界前列。

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代表论文

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