DNA甲基化在肿瘤诊断中

单基因诊断

SHOX2

多基因诊断模型

1，

2，

3，

血浆诊断生物标记物

让临床/科学家疑惑的是有些基因DNA甲基化可以在组织和血浆游离DNA中同时观测到，但有些基因的DNA甲基化只能在组织中检测到，无法在血浆游离DNA中观察到。是否意味着血浆中会选择性降解特定DNA片段或DNA甲基化片段。

1， 正常细胞中绝大多数CpG island都是hypomethylation的状态

肿瘤的亚型分类

急需解决的问题

Previous studies have shown that the promoters for somatic-tissue-specific genes are often CG poor and lack CpG islands (CGIs), in contrast to those for housekeeping genes, which are CG rich and predominantly contain CGIs ([Barrera et al., 2008](http://www.sciencedirect.com/science/article/pii/S0092867413004649#bib2); [Schug et al., 2005](http://www.sciencedirect.com/science/article/pii/S0092867413004649" \l "bib41)).

CGIs for promoter function by destabilizing nucleosomes and attracting proteins that create a transcriptionally permissive chromatin state. Silencing of CGI promoters is achieved through dense CpG methylation or polycomb recruitment.