# 1507882392(1)

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**癫痫基因检测报告**



Genetic Reports For Epilepsy

# 受检者信息

|  |  |
| --- | --- |
| **姓名：**#[name]# | **性别：** #[gender]# |
| **出生日期/年龄：#**[date\_of\_birth]# | **联系电话：**#[phone]# |
| **身份证号：**#[ID]# | **条码号：**#[GSID]# |
| **检测项目：** #[project]# | |
| **病理诊断：** #[diagnostic]# | |
| **治疗史：**#[treatment]# | |
| **家族史：**#[family]# | |

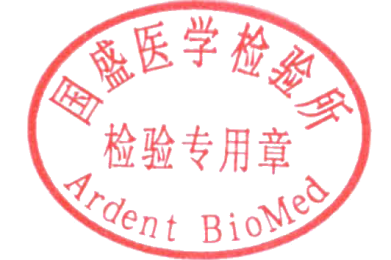
|  |  |  |
| --- | --- | --- |
| **样本类型：** #[type]# | **样本数量：**#[amount]# | **送检医生：**#[doctor]# |
| **采样日期：**#sampling\_date# | **收样日期：**#[collection\_date]# | **报告日期：**#[report\_date]# |
| **送检机构：**#[Inspection\_agencies]# | | |

**特别说明：**

1. 本报告仅对送检样本的此次检测结果负责。

2. 本报告属于个人隐私，本中心将严格保护被检者个人信息与检测结果不被泄露，请被检测人和相关人士认真阅读本报告和相关资料，并按照协议条款，对本报告及其相关资料自觉履行保密义务；因受检者个人原因出现的信息外泄，本公司不承担相应责任。

3. 由于科技不断发展，世界范围内数以万计的科学家正在夜以继日地致力于揭示基因和疾病的研究，我们会随时关注相关的研究进展，根据最新科研成果调整和丰富基因检测的内容。本检测只对检测结果的当前正确性负责并承诺检测服务的准确率保持在国际先进水平上。

4. 本公司承诺在当前科学技术条件下所有检测结果是真实的、有效的，基因检测结果的解释权归国盛医学检验所所有。

检测者：霍云龙 审核者：邱明

**TSC1、TSC2基因变异结果**

**1基因变异结果统计**

本次检测应用二代测序技术检测了与TSC1、TSC2基因相关位点。与正常人群基因库信息比对，根据ClinVar数据库分类，位点突变数目统计结果如下：

**表1：基因变异结果统计**

|  |  |  |  |
| --- | --- | --- | --- |
| **基因变异致病性分类（ClinVar数据库）** | **致病性**  **（Pathogenic）** | **可能致病**  **（Likely Pathogenic）** | **其他** |
| 检测总位点数 | #[pathogenic1]# | #[lp1]# | #[other1]# |
| 变异位点数 | #[pathogenic2]# | #[lp2]# | #[other2]# |

注：表格中致病性的分类是从严格的孟德尔遗传方式并采用ACMG推荐的五级分类系统。ACMC推荐序列变化可以分类为:

\*Pathogenic — this variant directly contributes to the development of disease. Some pathogenic variants may not be fully penetrant. In the case of recessive or X-linked conditions, a single pathogenic variant may not be sufficient to cause disease on its own. Additional evidence is not expected to alter the classification of this variant.

\*Likely pathogenic — this variant is very likely to contribute to the development of disease however, the scientific evidence is currently insufficient to prove this conclusively. Additional evidence is expected to confirm this assertion of pathogenicity, but we cannot fully rule out the possibility that new evidence may demonstrate that this variant has little or no clinical significance.

\*Uncertain significance — There is not enough information at this time to support a more definitive classification of this variant.

\*Likely benign — this variant is not expected to have a major effect on disease however, the scientific evidence is currently insufficient to prove this conclusively. Additional evidence is expected to confirm this assertion, but we cannot fully rule out the possibility that new evidence may demonstrate that this variant can contribute to disease.

\*Benign — this variant does not cause disease.

**2变异位点结果**

**表2：变异位点结果**

| **基因** | **位置** | **rs号** | **转录本号** | **碱基突变** | **氨基酸改变** | **人群分布率** | **致病等级(ClinVar)** | **基因型** | **Coverage** |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| #[FILLTABLE-change]# | a | b | c | d | r | f | g | h | i |

**3未变异的致病性位点结果展示**

**表3：未变异的致病性位点结果展示**

| **基因** | **位置** | **rs号** | **转录本号** | **碱基突变** | **氨基酸改变** | **人群分布率** | **致病等级(ClinVar)** | **基因型** | **Coverage** |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| #[FILLTABLE-DC]# |  |  |  |  |  |  |  |  |  |

注："-"表示无相关信息,Het表示杂合子,Hom表示纯合子,Wt表示野生型

* **小结：**

本次检测发现#[pathogenic2]#个致病性位点变异。检测到受检者携带了#[lp2]#个临床意义未明确的变异位点，其中#[rare]#个为罕见变异。

**4基因及相关疾病介绍**

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| --- |
| TSC1/2 |
| 1. 基因功能信息： TSC1和TSC2是常染色体基因,在体内作为抑癌基因广泛表达,表达产物Hamartin/TSC1和Tuberin/TSC2直接相互作用形成异二聚体(TSC1/TSC2) ，在mTOR信号通路中起到负调控的作用。TSC1/2复合能够整合上游的氨基酸、生长因子、能量、低氧和细胞因子等生长信号,转化为TSC1和TSC2不同位点的磷酸化状态。TSC2的GTP酶激活蛋白(GAP)结构域活化Rheb,从而抑制mTORC1(哺乳动物雷帕霉素靶蛋白复合物1)的活性。经由Rheb/mTORC1模式信号途径来调控细胞生长、分化及迁移并在胚胎发育及机体代谢过程中发挥重要的功能。 2. 基因变异与相关疾病：结节性硬化症(Tuberous Sclerosis Complex,TSC)是一种常染色体显性遗传性疾病。由肿瘤抑制基因TSC1和TSC2发生突变所致,不同突变导致的病症和严重程度都不同。由于TSC1/TSC2基因异常导致其转录产物功能的异常,从而影响了正常的细胞生成、分化和移行过程,病理上主要表现为细胞的形态、数量、位置以及结构排列异常而形成瘤样病变。临床表现为皮肤、神经系统、心脏、肾脏、眼底、肺脏等多脏器损害。该疾病表现出多种临床病症,如癫痫、自闭症、肾血管平滑肌瘤等。 3. 参考资料来源：[1]OMIM [2]Genetic Home Reference |

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