

生物医药合作项目开发

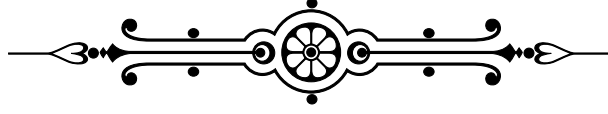
研究方向： 测试

委 托 人： _____

受 托 人： 杭州铂赛生物科技有限公司

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1 研究背景

1.1 相关研究

骨肉瘤病理生理机制涉及与骨形成相关的几种可能的疾病遗传驱动因素, 导致恶性进展和转移 (2022, Nature reviews. Disease primers, **IF:76.9**, Q1)¹。

1.2 相关概念

因果基因 (2009, Statistics Surveys, **IF:11**, Q1)²。

TWAS (2016, Nature Genetics, **IF:31.7**, Q1)³

PWAS (2020, Genome Biology, **IF:10.1**, Q1)⁴

1.3 思路

骨肉瘤 + 因果基因筛选 (联合 PWAS 和 TWAS) (可能筛选到线粒体失调相关)

涉及方法: - PWAS: GWAS + FUSION (2016, Nature Genetics, **IF:31.7**, Q1)³ <http://gusevlab.org/projects/fusion/> - TWAS: GWAS + S-PrediXcan (2018, Nature Communications, **IF:14.7**, Q1)⁵ <https://github.com/hakyimlab/MetaXcan> + FOCUS (2020, Human genetics, **IF:3.8**, Q2)⁶ <https://github.com/ChongWu-Biostat/FOGS>

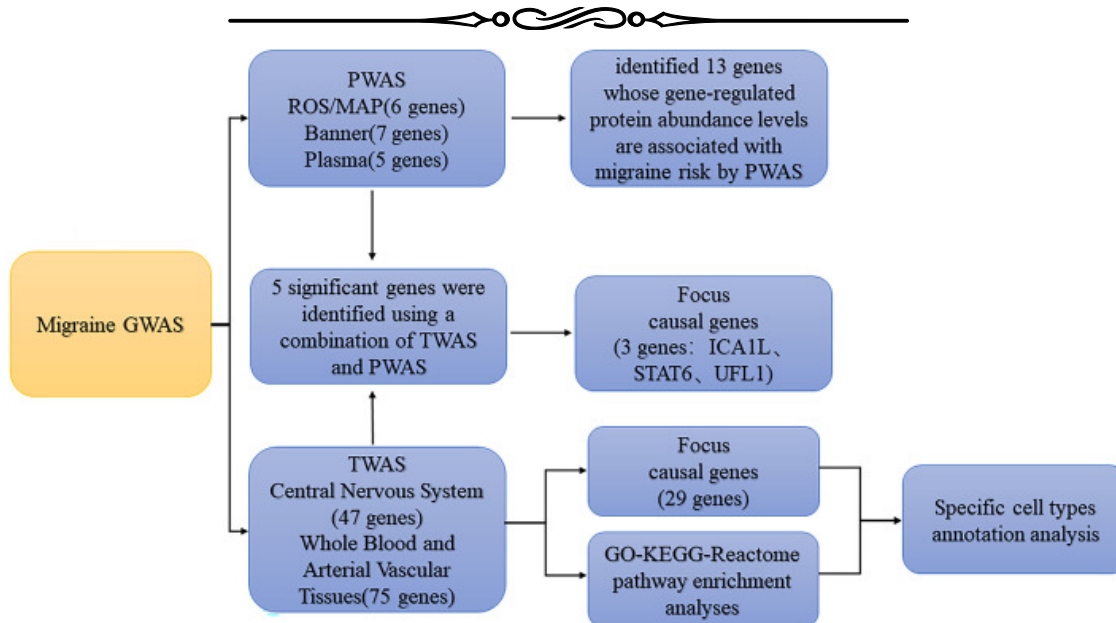


Figure 1: Example workflow

2 可行性

3 创新性

4 参考文献和数据集

Identifying causal genes for migraine by integrating the proteome and transcriptome (2023, The journal of headache and pain, **IF:7.3**, Q1)⁷

Reference

1. Beird, H. C. *et al.* Osteosarcoma. *Nature reviews. Disease primers* **8**, (2022).
2. Pearl, J. Causal inference in statistics: An overview. *Statistics Surveys* **3**, (2009).
3. Gusev, A. *et al.* Integrative approaches for large-scale transcriptome-wide association studies. *Nature Genetics* **48**, 245–252 (2016).
4. Brandes, N., Linial, N. & Linial, M. PWAS: Proteome-wide association study linking genes and phenotypes by functional variation in proteins. *Genome Biology* **21**, (2020).
5. Barbeira, A. N. *et al.* Exploring the phenotypic consequences of tissue specific gene expression variation inferred from gwas summary statistics. *Nature Communications* **9**, (2018).
6. Wu, C. & Pan, W. A powerful fine-mapping method for transcriptome-wide association studies. *Human genetics* **139**, 199–213 (2020).
7. Li, S.-J. *et al.* Identifying causal genes for migraine by integrating the proteome and transcriptome. *The journal of headache and pain* **24**, (2023).