The apa-sequencing Documentation

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Contents

Preface

This documentation is intended to describe the apa-sequencing[1] project.

Objective

Primary aldosteronism (PA) is the most common cause of secondary hypertension and is more common than primary hypertension in causing secondary diseases such as stroke, myocardial infarction, heart and renal failure. It is caused by inappropriately increased, partially autonomous synthesis and secretion of the steroid hormone aldosterone with consequent increased renal and intestinal sodium and water reabsorption and increased potassium secretion. The most common causes are a unilateral benign adrenal tumour (aldosterone-producing adenoma, APA) or bilateral aldosteronism. In recent years, the molecular mechanisms leading to autonomous aldosterone production have become the focus of increasing scientific attention. It has been shown that approximately 95% of all APAs have somatic mutations in known disease genes, mainly affecting ion channels and transporters: About 40% have KCNJ5 mutations; CACNA1D is the second most commonly affected gene (about 20%). Other less common somatic mutations include mutations in the ATPases ATP1A1 and ATP2B3 and the gene encoding b-catenin, CTNNB1.

Methods

Results

Conclusion

Outlook

[1] https://github.com/scholl-lab/apa-sequencing