INPLASY PROTOCOL

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Conflicts of interest:
None declared.

Mutations and clinical characteristics of dRTA caused by SLC4A1 mutations

Yang, MG[1]; Liao, L[2].

Review question / Objective: **To explore the comparisons of heredity and clinical characteristics between Asian and Non-Asian.**
Condition being studied: **The SLC4A1 gene is one of the bicarbonate anion transporters of the solute carrier family 4 (SLC4) gene family. Mutations in SCL4A1 may cause distal renal tubular acidosis (dRTA). Our understanding of dRTA caused by SLC4A1 mutations is limited by low incidence and phenotypic variability, and mutations in SLC4A1 have not been systematicly documented. However, diagnosis and treatment are often delayed due to clinical variability of the disease, and hereditary kidney disease compromises the quality of life of patients. In this sysmetic review, we analyzed the genetic defects in SLC4A1 and clinical phenotypes of the patients to facilitate the diagnosis and treatment of dRTA with SLC4A1 mutations.**

INPLASY registration number: This protocol was registered with the International Platform of Registered Systematic Review and Meta-Analysis Protocols (INPLASY) on 08 December 2022 and was last updated on 08 December 2022 (registration number INPLASY2022120031).

INTRODUCTION

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Condition being studied: **The SLC4A1 gene is one of the bicarbonate anion transporters of the solute carrier family 4 (SLC4) gene family. Mutations in SCL4A1 may cause distal renal tubular acidosis (dRTA). Our understanding of dRTA caused by SLC4A1 mutations is limited by low incidence and phenotypic variability, and mutations in SLC4A1 have not been systematicly documented. However, diagnosis and treatment are often delayed due to clinical variability of the disease, and hereditary kidney disease compromises the**

quality of life of patients. In this sysmetic review, we analyzed the genetic defects in SLC4A1 and clinical phenotypes of the patients to facilitate the diagnosis and treatment of dRTA with SLC4A1 mutations.

METHODS

Participant or population: Patients were diagnosed as dRTA with SLC4A1 mutations.

Intervention: Effect of different races on clinical characteristics of patients with dRTA.

Comparator: Aian patients and non-Asian patients.

Study designs to be included: Observational study.

Eligibility criteria: (1) patients are diagnosed as dRTA. (2) mutations in SLC4A1 are confirmed using molecular genetic techniques. (3) clinical data of patients are described. The articles involving a series of patients but without detailed descriptions are excluded.

Information sources: PubMed, Embase, Web of Science, the China National Knowledge Infrastructure, and Wanfang.

Main outcome(s): To explore the comparisons of heredity and clinical characteristics between Asian and Non-Asian, which include the incidence of nephrocalcinosis, kidney stones, developmental disorders, hematological abnormalities, renal dysfunction, muscle weakness and gastrointestinal symptoms.

Quality assessment / Risk of bias analysis: the Newcastle-Ottawa Scale, NOS.

Strategy of data synthesis: The epidemiological and clinical characteristics, and laboratory indexes of patients will be described utilizing simple summary statistics. Mann-Whitney U test and was t-test are used to analyze the data. The significance level is set as $p < 0.05$. Statistical analysis is performed using the Statistical Package for the Social Sciences version 26 for Windows (SPSS).

Subgroup analysis: Subgroup analyses were performed according to gender and ethnicity.

Sensitivity analysis: Excluding literatures one by one: Effect size combination was carried out after removing each included study one by one, and effect size combination was carried out after the inclusion and exclusion criteria were changed or a certain type of literature was removed.

Country(ies) involved: China.

Keywords: distal renal tubular acidosis; SLC4A1; metabolic acidosis; mutation; clinical characteristics.

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