MOLECULAR AND PHENOTYPIC PROFILE OF ALSTROM SYNDROME IN CHINESE PATIENTS

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INTRODUCTION

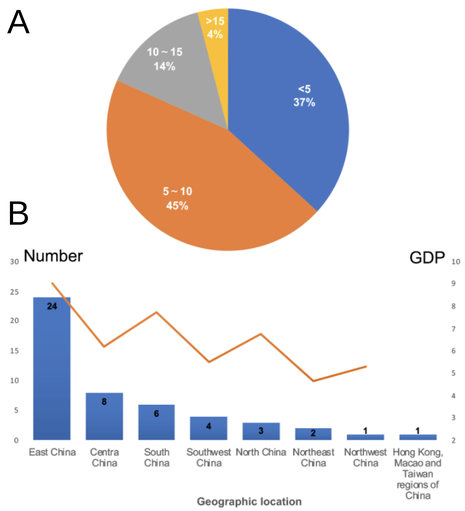
Alstrom syndrome, inherited in autosomal-recessive manner, is a complex multi-system disease including obesity, sensorineural hearing loss, retinal dystrophy, cardiomyopathy, type 2 diabetes mellitus, and multiple organ fibrosis. ALMS1 is the defective protein of Alstrom syndrome. Large cohorts of Alstrom syndrome lack around the world, especially in East Asia.

AIM

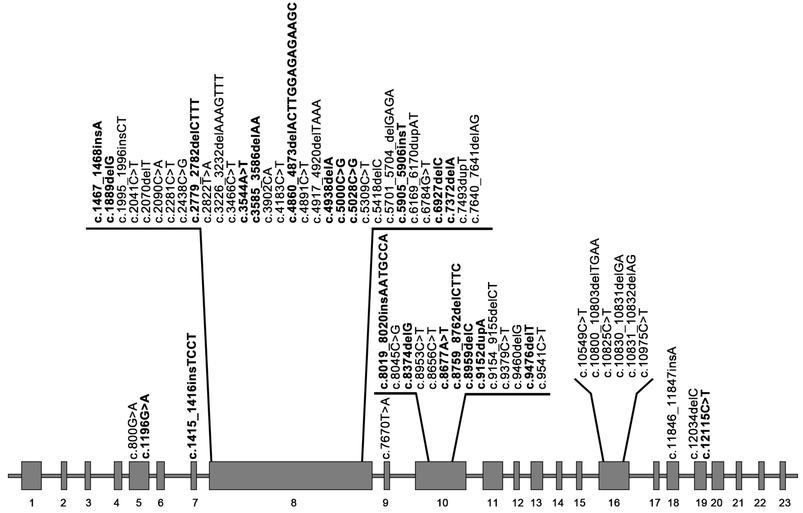
1. Expanding the *ALMS1* gene genetic and phenotypic spectrum
2. Enriching Asian data about Alstrom syndrome
3. Providing new insights into understanding of ALMS

RESULTS

1. *Most of the patients with Alstrom syndrome in our cohort were under 10 age years old*
2. *Most of the patients in China were from East China, Centra China and South China.*
3. *61 different ALMS1 variants (59 truncating and 2 exon deletion) in 50 patients from 47 different families were confirmed, including 23 novel variants.*
4. *The variant c.2090C>A was the most frequency variants in Chinese cohort.*
5. *Retinal dystrophy was observed in all patients. Obesity and hepatic symptom were shown in over 50% of the patients.*



Basic information of the cohort. A. Numbers of patients and real GDP per capita in different areas in China. B. Age distribution of 50 patients with Alstrom syndrome. C, D. Height for age of the patients in boys and girls (0-18y) respectively.



ALMS1 variants identified in this study. Novel variants are shown in bold.

METHOD

50 patients were included in this study, aged from 0.47 years to 21.66 years old. Detail phenotypic data and genetic data were obtained from all affected individuals. Truncated mutations were confirmed in all patients through genetic sequencing with Alstrom syndrome. And all Chinese patients reported previously were reviewed.

CONCLUSIONS

This is the largest cohort of Chinese patients with Alstrom syndrome and it is the youngest cohort in the world. This study enriched the spectrum of genotypes and phenotypes.

REFERENCES

1. Marshall, J.D., et al., Alström Syndrome: Mutation Spectrum of ALMS1. Hum Mutat, 2015. 36(7): p. 660-8.

2. Brofferio, A., et al., Characteristics of cardiomyopathy in Alström syndrome: Prospective single-center data on 38 patients. Mol Genet Metab, 2017. 121(4): p. 336-343.

3. Tahani, N., et al., Consensus clinical management guidelines for Alström syndrome. Orphanet J Rare Dis, 2020. 15(1): p. 253.

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