MOLECULAR AND PHENOTYPIC PROFILE OF ALSTROM SYNDROME IN CHINESE PATIENTS

Q. ZHANG1, Y. DING1, B. FENG, Y. TANG1 and X. WANG1
1. Department of Endocrinology and Metabolism, Shanghai Children’s Medical Center, School of Medicine, Shanghai Jiao Tong University, Shanghai, China

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

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.

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 frequent variants in Chinese cohort.
5. Retinal dystrophy was observed in all patients. Obesity and hepatic symptom were shown in over 50% of the patients.

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

ACKNOWLEDGEMENTS
This work was supported by National Nature Science Foundation of China (81900722) and Pudong New Area Science and Technology Development Fund (Grant PKJ2018-046).

CONTACT INFORMATION
Q. ZHANG, zhangqw@sjtu.edu.cn
X. WANG, wangxiumin1019@126.com