P198

Insights into the Cardiac Phenotype in Kabuki Syndrome

Leen Khalife,MD; Sidney Brown,BSC; Natalie Formica,BS; Olaf Bodamer,MD,PhD

Boston Children's Hospital/Harvard Medical School, Boston, MA

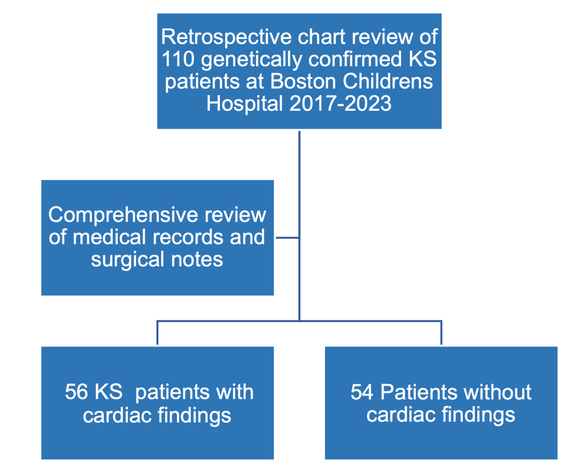
INTRODUCTION

- Kabuki Syndrome (KS) is a rare and complex heterogeneous congenital malformation condition characterized by a characteristic facial gestalt, variable intellectual disability, hypotonia, postnatal growth deficiency, immune deficiencies and congenital cardiac anomalies.
- KS is due to heterogenic variations in *KMT2D* (OMIM # 147920, 90%+ patients) and heterozygous or hemizygous variations in X-linked *KDM6A* (OMIM # 300867, 5-10%).
- There is a well-established correlation between KS and Congenital Heart Defects (CHD) with an estimated prevalence of 28% to 80%.
- While previous studies have examined patterns of cardiological findings, limitations included a lack of molecular confirmation of KS and/or small cohort sizes.

AIM

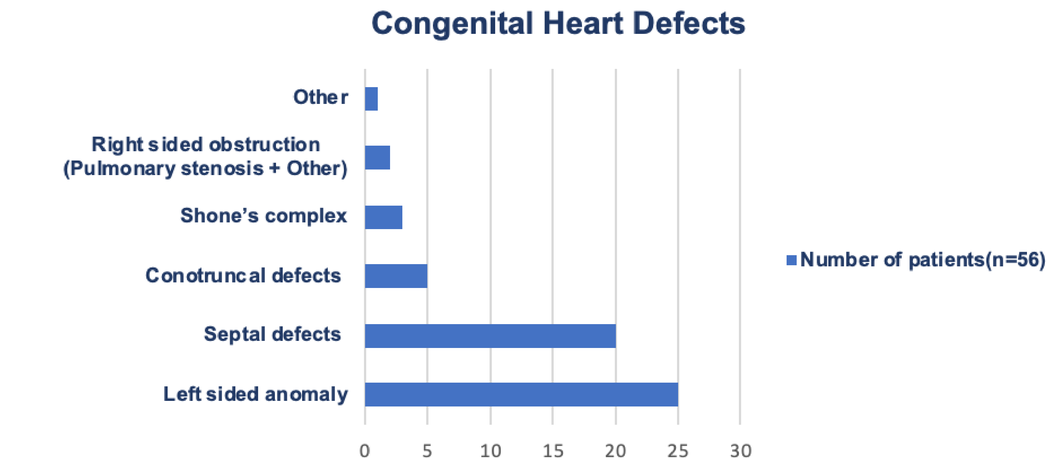
Delineate the spectrum of cardiac phenotypes and validate earlier observation in the literature in the largest set of molecularly confirmed KS patients in a single tertiary care center.

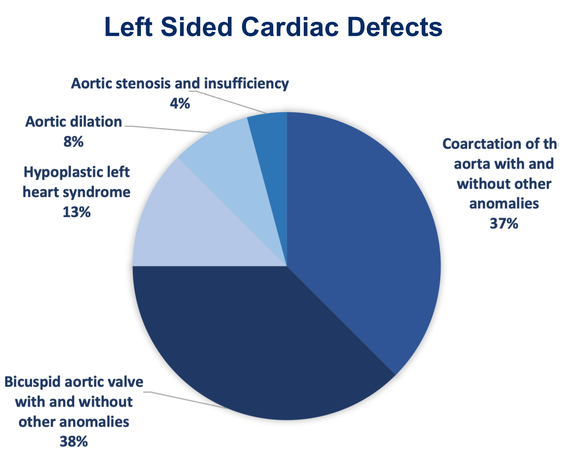
METHODS

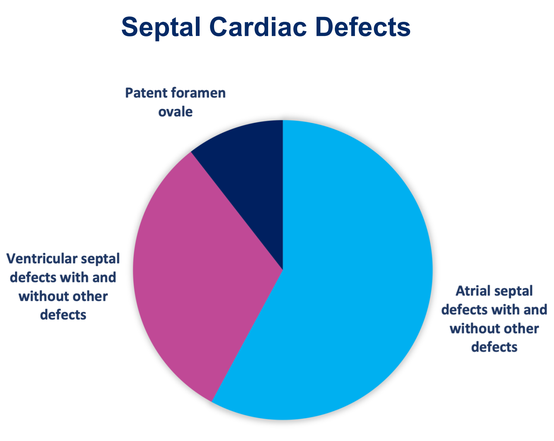


RESULTS

54 X *KMT2D*
2 X *KDM6A*







CONCLUSION

- This study examined the cardiac phenotype in the largest sample of genetically confirmed KS patients.
- The cardiac manifestation in KS displayed varying degrees of complexity, with more than half of the patients demonstrating multiple concurrent anatomical defects.
- Consistent with previous studies, left sided heart anomalies and septal defects were among the most prevalent findings.
- This study underscores the importance of cardiac assessment for individuals diagnosed with KS. It emphasizes the importance of conducting an echocardiogram, including aortic arch visualization, at the time of diagnosis and maintaining regular follow-up and surveillance for those with identified CHD .

REFRENCES

- Yoon, J. K., Ahn, K. J., Kwon, B. S., Kim, G. B., Bae, E. J., Noh, C. I., & Ko, J. M. (2015). The strong association of left-side heart anomalies with Kabuki syndrome. Clinical and Experimental Pediatrics, 58(7), 256–262. https://doi.org/10.3345/kjp.2015.58.7.256
- Conrey, R., Tume, S., Bonilla-Ramirez, C., Lalani, S., McKenzie, D., Marc, & ers. (2021). Kabuki-Syndrome and Congenital Heart Disease—A Twenty-Year Institutional Experience. Congenital Heart Disease, 16(2), 171–181. https://doi.org/10.32604/CHD.2021.014409

CONTACT

Name: Leen Khalife
Organization: Boston Children's Hospital
Email: Leen.khalife@childrens.harvard.edu
Financial Disclosures : None