Introduction:
Multiple mutations in the SCN5A gene is rare (3%). We describe a Chinese family with Brugada syndrome (BrS). The proband carries both the SCN5A p.A227V and the p.R1629X mutation.

Methods:
PN (proband) presented with syncope at the age of 11. His DNA was analyzed for SCN5A mutations. Clinical evaluation and cascade genetic screening was performed for the entire family.

Results:
PN had spontaneous type 1 coved ST-segment elevation and prolonged PR and QRS intervals on ECG. His QRS duration showed progressive prolongation during exercise prior to VT onset. PN’s father, NC (age 56), is heterozygous for p.A226V and phenotypically normal. LM (age 54), PN’s mother, is heterozygous for p.R1629X. Her PR interval and QRS duration are prolonged. Both parents are asymptomatic. PH (age 16) is PN’s sister, she is heterozygous for p.A226V. CN (age 24), is PN’s brother, does not carry either mutation. Both siblings are phenotypically normal and are asymptomatic.

Conclusion:
Compound heterozygosity in PN resulted a more severe phenotype. The radical truncated mutation p.R1629X is expected to confer a more severe phenotype. However, both parents and PH remains asymptomatic. Additional genetic and environmental factors may play a role in disease manifestation in BrS.

Keywords: Brugada syndrome, SCN5A