

CARDIAC CAUSES OF SUDDEN UNEXPLAINED INFANT DEATH

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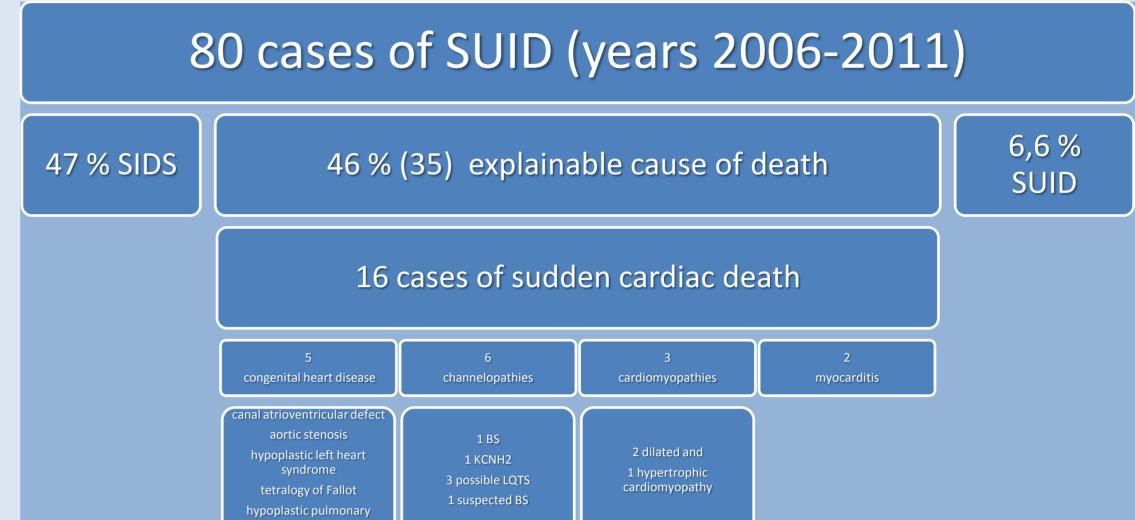
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OBJECTIVES: To evaluate cardiac causes of sudden unexplained infant death (SUID) in our county with a multidisciplinary approach.

MATERIAL AND METHODS: Post-mortem investigations followed the San Diego classification. DNA from blood samples was preserved for genetic studies in selected cases (KCNQ1, KCNH2 and SCN5A genes). First degree relatives underwent cardiological evaluation with ECG, echocardiography and exercise testing (optional Holter-ECG monitoring).

RESULTS:

- 80 cases of SUID from 2006 to 2011 were reviewed.
- Thirty-six cases (47%) had been diagnosed as sudden infant death syndrome (SIDS),
 5 (6.6%) as undetermined and 35 (46%) had an explainable cause of death.
- Cardiac diseases (16) are the second cause of explainable death (52% are infectious diseases) and included: **congenital heart disease** (canal atrioventricular defect, aortic stenosis, hypoplastic left heart syndrome, tetralogy of Fallot, and hypoplastic



pulmonary artery), **channelopathies** (6: 1 carriving a SCN5A mutation causing Brugada syndrome-BS, 1 carriving 2 mutations in KCNH2 causing long QT syndrome-LQTS, 3 possible LQTS with negative genetic study, 1 suspected BS with negative genetic study), **cardiomyopathies** (2 dilated and 1 hypertrophic cardiomyopathy) and myocarditis in the context of systemic infection (2).

- Males predominate in all groups (12/16).
- Congenital heart diseases have a significant earlier presentation (22 days) than channelopathies-myocarditis (4 months) and cardiomyopathies (7 months).



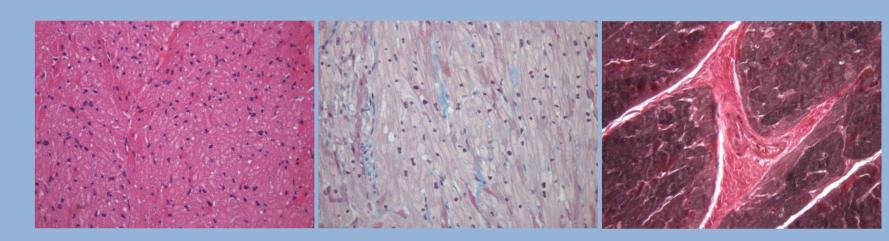
Sudden cardiac death in a 7-day-old male infant: **severe congenital malformation** with hypoplasia of the right branch of the pulmonary artery (*) with right pulmonary hypoplasia and interventricular and interventricular communication.



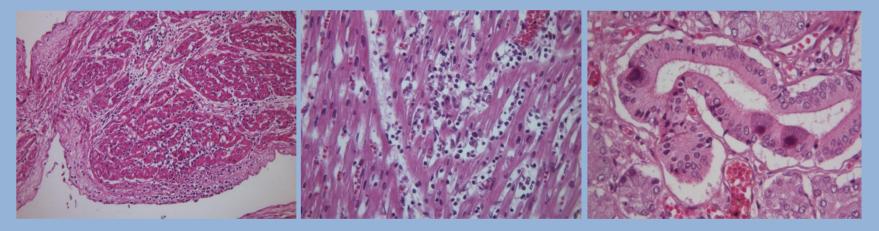
Sudden cardiac death in a 7-month-old male infant with **hypoplastic left heart syndrome** with aortic coarctation, underdeveloped mitral valve and left ventricle, hypoplastic aortic valve (bicuspid) and compensatory right heart dilatation



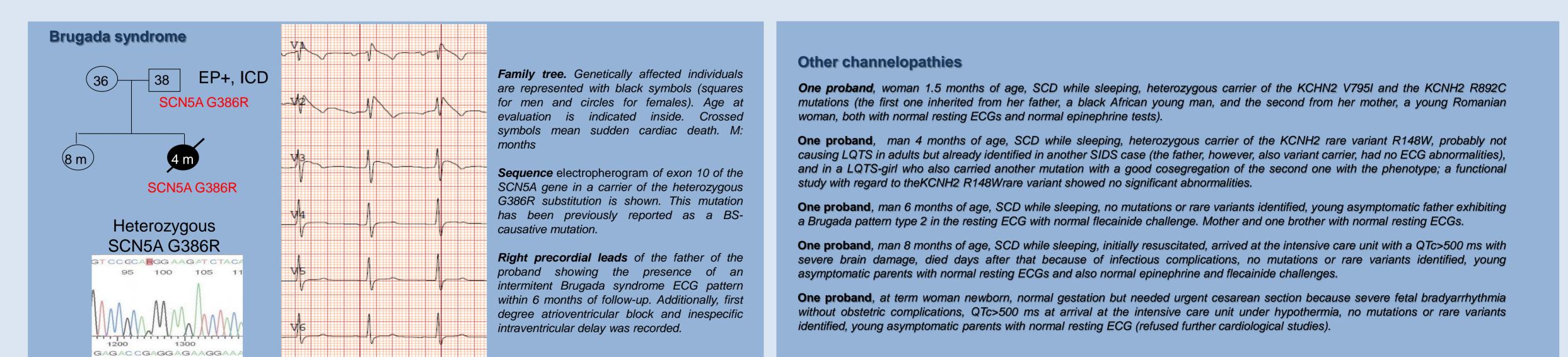
Sudden cardiac death in a 7-month-old male infant with **dilated cardiomyopathy**. Heart weight: 79 grs (average value per body weight: 39-44 g (range 27-30;58-64 grs). Thickness left ventricle: 0.5 cm, interventricular septum: 0.5 cm and right ventricle: 0.1 cm. Patent foramen ovale. Biventricular parietal thinning with dilated left ventricular cavity (diameter 2 cm). Subendocardial fibroelastosis.



Sudden cardiac death in a 9-month-old male infant with **hypertrophic cardiomyopathy** due to **long-chain fatty acid oxidation defects**. Histological specimen from the left ventricle demonstrating fat accumulation, HE (left), Masson Trichrome (center) and Osmium tetroxide (right) staining. Genetic study: carnitine-acylcarnitine translocase deficiency (homozygous mutations c.59G> A)



Sudden cardiac death in a 6-month-old male infant with myocarditis. The microscopic analysis confirmed multiples inflammatory infiltrates with myocardial necrosis in all the sections studied (right and central images). Typical cytomegalovirus-induced inclusion bodies (left) was observed in the ductal cells of the submandibular gland.



DISCUSSION:

Sudden cardiac death is a common cause of SUID, follow SIDS and infectious diseases. Congenital heart diseases can not be diagnosed during the birth hospitalization because most are primarily ductal dependent defects. Our protocol allows to diagnosis channelopathies and avoid misdiagnosis of unclassified sudden infant death in this cases. Unfortunately, this multidisciplinary study still depends upon research grants and has not been assumed by public institutions.

CONCLUSIONS:

Cardiac multidisciplinary approach is pivotal to exclude channelopathies from SIDS series and may yield substantial benefit to the society as long as early diagnosis and treatment can be implemented in the affected living relatives.

