

MP4-1

Early Detection of Sudden Infant Death Syndrome: Electrocardiogram, Genetic Analysis and Familial Assessment.

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Background. Arrhythmogenic diseases have been proven to cause infant sudden death. Some of them can be diagnosed performing a 12-lead ECG at birth.

Objective. We pretend perform an ECG in all births in order to identify the percentage of any malignant electrical alteration in neonatal population.

Methods. We have performed a prospective analysis of ECG performed at 24 to 72 hours of life to new-borns of a single centre. Of those which QT interval was above normal, saliva was collected for DNA genetic analysis using a gene panel related to sudden death using ultrasequencing technology. Clinical and genetic analysis of the family members has been performed accordingly.

Results. During the first year of the project we have performed 600 ECG in new-borns (51% males). We have identified 14 cases of prolongation of the QTc above 470ms and ECGs of the first-degree family

members has been performed. In 7 of them (QTc below 490ms) normalization of the ECG has been observed along the first 6 months of life and no family members have been detected abnormal. In the remaining 7 cases with persistent QTc interval above 490 ms., a genetic analysis has been performed. In 6, family members ECG were abnormal. In 5, a rare genetic variant has been described (one of them, de novo).

Conclusions. Neonatal ECG screening allows early detection of potentially lethal cardiac diseases. Genetic study helps identifying the underlying mechanism of the disease. Family cascade study is mandatory in order to detect other disease carriers.