G80 A Case of Sudden Death in a 3-Year-Old Infant With Prader Willi Syndrome

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A case of sudden death of a 3-year-old infant affected by Prader Willi Syndrome is presented. The aim of the paper is to analyze the macro-microscopic findings in Prader Willi Syndrome and to focalize the forensic implication in sudden death cases.

To the best of our knowledge, this is a rare case of sudden cardiac death in infant with Prader Willi Syndrome. This datum is confirmed by means of histological study of cardiac tissues. It would be an important contribute to the scientific community for the diagnosis of the cause of death in Prader Willi Syndrome.

The Prader Willi Syndrome has a variable prevalence of 1:10000 – 25000.

The syndrome is a rare genetic disorder caused by a chromosomal aberration. Most common a deletion on chromosome 15q11-13 in the portion inherited from the father, less frequently the child has two chromosomes 15 from the mother and none from the father, rarely there is an imprinting mutation on chromosome 15q11-q13. The Prader Willi Syndrome is characterized from a hypothalamic dysfunction that leads to hyperfagia and obesity and has secondary consequences of diabetes, heart disease, stroke and sleep apnea. It is also characterized by severe muscular hypotonia, short stature, cryptorchidism, learning disabilities, mental retardation.

We present a sudden death case in a 3 years old young male, affected by Prader Willi Syndrome diagnosed by means of DNA methylation. The infant was referred apparently unconsciousness by his mother to the Emergency Area. Pulse was absent, he was breathless and in fixed mydriasis. The cardiopulmonary resuscitation was unsuccessful. In the history, mother referred that her son suddenly collapsed while he was on bed.

The external examination was performed showing an uninhabited scrotum, a severe hypo tonic muscular body mass and an excessive accumulation of fat. At the post mortem examination the skull cup was uninjured, the brain was congested and edematous with convolutions reduced (microgyria) in correspondence of parietal and occipital lobes, but normal in size, volume and weight. It was fixed in formalin and later it was sectioned with coronal cuts according to the Pitres technique, showing hypoplasia of olive of medulla oblongata and haemorrhage on periventricular surface. Cervical and thoracic organs were removed "en block" according to the Ghon's technique, appeared edematous and congested but anatomically normal; tracheobronchial tree and lungs, were unremarkable except for white fluid in upper respiratory tract. Heart was fixed in formalin and a dissected according to "Four – Chamber" method was completed. Cardiac size was normal, with conical shape, the color of fresh subepicardial myocardium was reddish-brownish. Macroscopic study (cut in cross-section 3 mm intervals) of coronary arteries was unremarkable. Left ventricular thickness, measured 2 cm below mitral anulus, was cm 1; ventricular septum 1.2 cm; right ventricular thickness 0.4 cm. Histological cardiac findings were represented by spotty area of fibrosis; myocells showed eosinophilic cross-bands consisting of segments of hyper contracted or coagulated sarcomeres, to a total disruption of myofibrils and cells with granular aspect. In particular, the contraction band necrosis were variously distributed in multiple foci, formed by few myocells. Sarcomeres appear shorter than their normal length and this finding was associated with marked thickening of the Z-lines. Granular destruction of myofibrils were also associated with a paradiscal lesion (paradiscal contraction band) without rhexis of the myofibrillar apparatus. Adjacent normal myocells show a typical "wavy" disposition, possibly induced by the hyper contracted myocells. Myocardial fibers appear also stretched and broken. (A quantitative morphometric analysis has been conducted). In the lung mild pulmonary oedema was observed; a limited bronchial phlogistic infiltration, and large areas of atelectasia were also observed.

The histological findings lead to the definition of a cardiac death with a typical picture of contraction band necrosis (CBN). The cardiac findings should be judged sufficient to explain the cause of death. Pulmonary hypoxic alteration is frequently reported as primary cause of death in PWS cases. The chronic hypoxic stimuli should trigger a fatal cardiac arrhythmia, as demonstrated by typical myocardial damage (Contraction Band Necrosis).

In conclusion, this case contributes to a better definition of morphological findings in sudden death related to Prader Willi Syndrome.

Prader Willi Syndrome, Sudden Cardiac Death, Histological Findings

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