G36 Sudden Death of a 17-Year-Old Boy Due to Suspected Williams Syndrome - A Case Report

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After attending this presentation attendees will be well acquainted with the external and internal findings/characteristics of the rare genetic syndrome Williams syndrome which can cause sudden death in young population.

The presentation will impact the forensic community by the fact that every case of sudden death must be scrutinized carefully both by reveling all the circumstances of the case with collecting relevant heteroanamnestic data and need to perform thorough examination of the body (external and internal). Finally, in order not to have only suspicion on some genetic disorder such as Williams syndrome, but to confirm it with the highest certainty, it is necessary to assign the task to develop genetic tests like FISH. Sudden and unexpected death is one of the frequent problems in common medicolegal practice, which attracts special attention when it pertains young and previously apparently healthy individuals. One of the potential causes of such death could rarely be Williams syndrome (WS) due to specific cardiovascular abnormalities. This syndrome was initially described by Williams et al. in 1961, and Beuren et al. explained the phenotype. WS is a sporadic genetic syndrome, with an estimated incidence of 1 in 20.000 live births, caused by a deletion of elastin gene and other contiguous genes at chromosome 7, with variable phenotypic expression, associated with dysmorphic facies, neurological manifestations, idiopathic hypercalcemia, and cardiovascular features, particularly supravalvular aortic stenosis. Namely, more than 90% of the patients with WS exhibit a submicroscopic deletion spanning at least 114 kb, at 7q11.23. Hemizygosity of the elastin gene could account for all connective tissue, especially the vascular, abnormalities seen in WS.

The case concerns the 17-year-old boy who was playing with his brother in the yard of their family house during wintertime, when he suddenly fell down to the snow and died shortly after. Hetero-anamnestically, his mother stated that in his childhood, the boy suffered from abdominal pains periodically, denying however, any illness diagnosed by the physicians. In addition, she mentioned his constant problems with learning and relationship with his friends as well (slight mental retardation and nervousness). Since the boy had not been medically examined, no other clinical data could be obtained. At the autopsy, external examination revealed elfin face with short and slightly upturned nose, long filtrum and very bad dental condition – black-greenish coloration of the crowns of the front teeth. There were no signs of mechanical injuries on the body. Internal examination showed severe narrowing (the circumference 4 cm) of the ascending aorta, some 4 cm above the semilunar valves (supravalvular aortic stenosis) with mild enlargement of the heart, weighted 340 grams and significant thickening of the left ventricular myocardium, measured 2,3 cm. With exception of hypertrophy of the myocardial fibers, the histological findings disclosed no pathological abnormalities in all other organs. Toxicological screening was negative. Primarily based upon the internal findings along with case circumstances, in the autopsy record it was inferred that death of the teenager was of natural manner, the most probably due to inherited cardiovascular abnormalities (narrowing of the aorta and hypertrophy of the left ventricular myocardium). Regardless the fact that the case had been solved concerning manner and cause of death, yet the forensic pathologists posed a question to themselves – what might be the origin of such peculiar combination of external and internal autopsy findings. The attention was focused on his specific facial appearance together with the cardiovascular status and obtained heteroanamnestic data, which raised a suspicion on Williams syndrome.

Since WS is a genetic disorder, besides the above mentioned clinical manifesations and morphological findings, for definite diagnosis it is necessary to perform specific genetic analysis - the FISH test, which is a type of specialized chromosome analysis utilizing specially prepared elastin probes. Unfortunately, at the moment, neither the Institute of Forensic Medicine in Novi Sad, nor that in Belgrade is equipped for such test.

Williams Syndrome, Sudden Death, FISH Test

 ** Presenting Author*