



G83 Unexpected Death of 24-Year-Old Male With a Phenotype Strongly Suggestive of Lujan-Fryns Syndrome

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After attending this presentation, attendees will be acquainted with features of a rare genetic disorder which may be of great interest in common forensic practice, but at the same time is very difficult to be recognized.

This presentation will impact the forensic community demonstrating that in certain cases sudden and unexpected death could be attributed to peculiar genetic disorder which has not been described until now in forensic literature.

The presented case concerns unexpected death of a 24-year-old male shortly after his discharge from the hospital where he was admitted due to severe psychiatric behavioral disorder. He had been treated in the course of three months with the following final diagnoses: moderate mental retardation, non-organic non-specified psychosis, and significant behavioral disorder which needs special care and treatment. He was sent home in the phase of psychiatric symptoms' remission. Heteroanamnestic data obtained from the family members revealed his cognitive and behavioral impairment many years before hospitalization; in addition, they described the physical status of the young man two days after his hospital discharge-frequent vomiting, severe abdominal pain, fever and exhaustion. Thus, he refused to eat and *ante finem* he even couldn't open his mouth. Together with other family members, his mother tried to help him by giving food and water, but all the efforts were in vain – he died in his bed. The police report confirmed all statements given by the deceased's family.

The external examination showed distinct facial dysmorphism (elongated narrow face, prominent forehead, long nose, maxillary hypoplasia, and small mandible), as well as marfanoid stature with long slender extremities. The internal examination disclosed aortic narrowing (circumference of 4.5 cm at the valves level), right aortic arch, low heart weight (240 g) with normal thickness of the left and right ventricle wall. The large intestine was widened, tensed, and full of gases and liquefied feces. No other pathological abnormalities were noticed in all other examined organs. Microscopically, slight congestion and edema of all tissues were found; specific staining for elastic fibers (Verhoeff method) showed rupture of elastic lamina and scarcity of elastic fibers with cystic degeneration of aortic media.

The postmortem toxicological screening detected the presence of clozapine and fluphenazine (neuroleptics) in the blood and bile, as well as clozapine and midazolam (benzodiazepine) in the stomach content. These findings fitted well the data in the medical records informing that the man received depot intramuscular injection of fluphenazine decanoate (25 mg) three days before his release and was instructed to continue with his therapy at home by taking clozapine (50 mg three times a day), lorazepam (2.5 mg three times a day) and midazolam (15 mg when needed at night). In the conclusion of the autopsy protocol, the cause of death was attributed to paralytic ileus due to antipsychotic therapy applied.

On the basis of the case circumstances, collected medical records, heteroanamnestic data and the autopsy findings - including peculiar facial appearance along with both macroscopical and microscopical cardiovascular features (narrowing of aortic root and ascending aorta and mediocystic degeneration of aortic wall), there was a strong suspicion on the Lujan-Fryns syndrome. In the available forensic literature the case of the Lujan-Fryns syndrome with fatal outcome is not found.

The Lujan-Fryns syndrome is defined by X chromosome-linked mild to moderate mental retardation, distinct facial dysmorphism (long narrow face, prominent forehead, long nose, maxillary hypoplasia, and small mandible), marfanoid stature with long slender extremities and behavioral problems. The genetic defect is not known; therefore the diagnosis is based on the presence of the clinical manifestations.

Lujan-Fryns Syndrome, Paralytic Ileus, Antipsychotics