

Sudden unexpected postnatal collapse (SUPC): analysis of two neonatal deaths in the same family

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Objective. In our study we focus on 2 SUPC-attributed neonatal deaths which occurred in the same family.

SUPC is a rare, difficult to identify event, so it is essential to draw the scientific community's attention to this condition in order to reach a correct diagnosis through a multidisciplinary approach.

Furthermore, there is currently no relevant scientific data which links SUPC to specific genetic disorders.

Materials and Methods. Our first case regards a full-term newborn with a 10-minute from birth 10-point Apgar score who, at 24-hours of age, suddenly developed severe respiratory distress associated with bradycardia and acidosis. Despite all resuscitation efforts, the baby did not survive.

One year later, her two-day old sister also died, in very similar circumstances.

In both cases, an autopsy and all relevant laboratory investigations were carried out.

Results. On the basis of such tests, the first baby's cause of death was determined to be SUPC.

In addition, when searching for significant pathogenic mutations for cardiomyopathies, a gene variant in the NKX2-5 gene of uncertain pathogenic significance was identified.

Conclusions. The two cases of SUPC that have occurred in this family call for the continuation and in-depth genetic analysis of the parents as well; this could make it possible to identify a possible correlation, as yet undocumented, between SUPC and gene mutations.