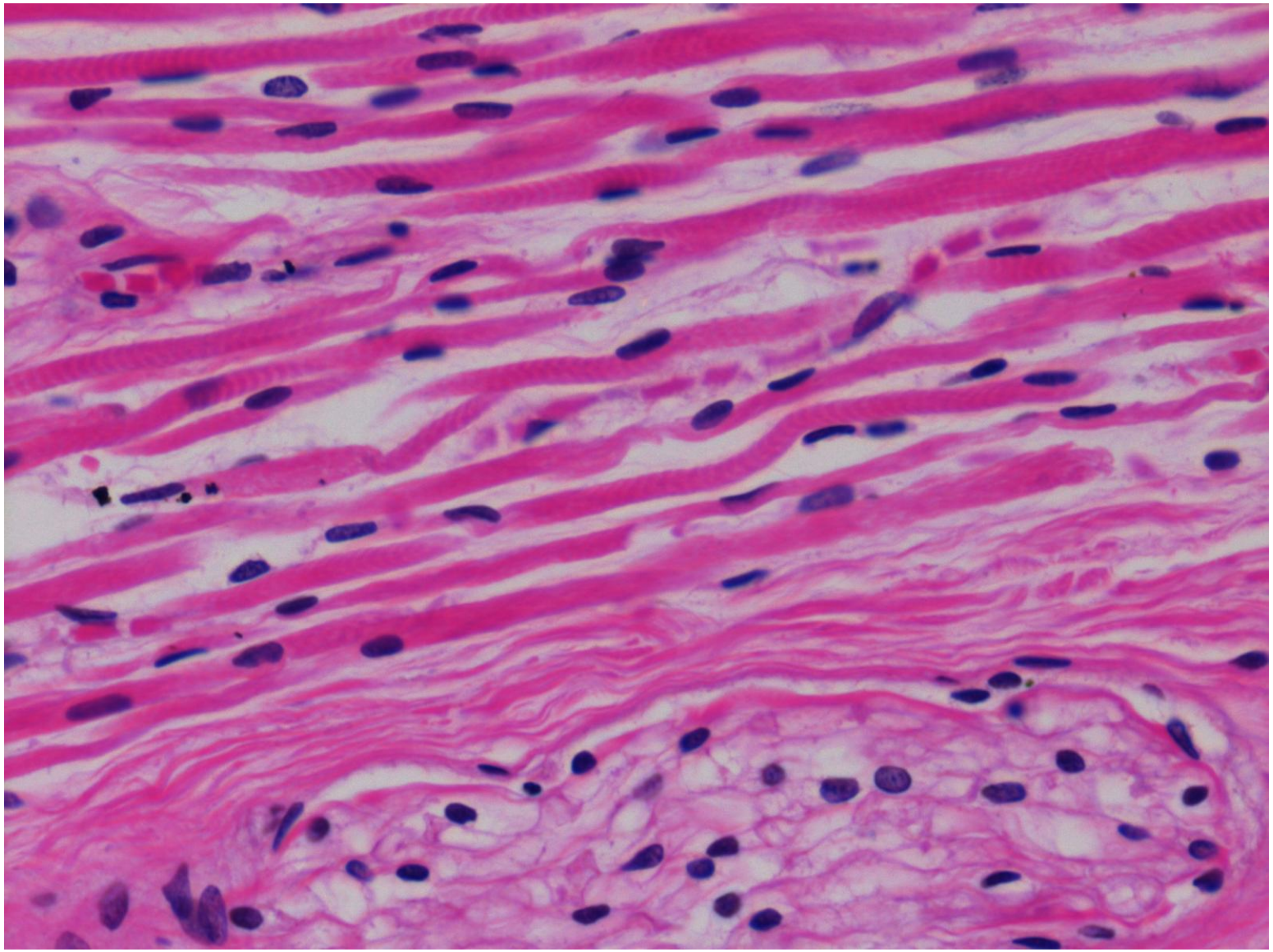
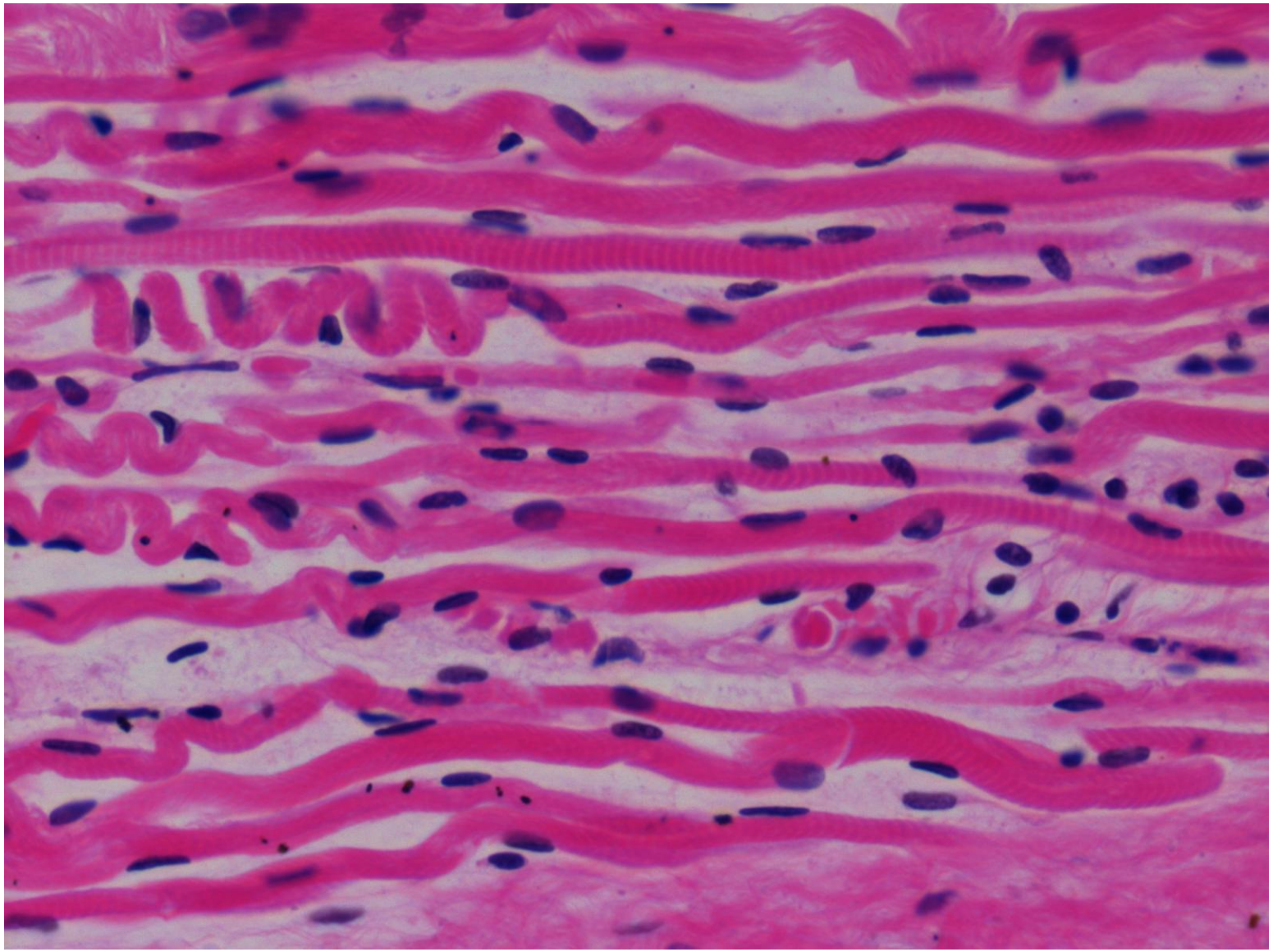
Congenital myopathy with fiber-type disproportion

Congenital myopathy with fiber-type disproportion characterized by type 1 fiber hypotrophy is associated with muscle weakness in the neonate, and has a non-progressive course. In one fourth of the patients, severe muscle weakness is seen at birth, and death follows in infancy. Here, a case of congenital myopathy with fiber-type disproportion who died soon after birth is presented. Others appear mild and non-progressive muscle hypotonia and weakness by age 1. Motor skills such as standing and walking may be delayed, but many children eventually learn to walk.

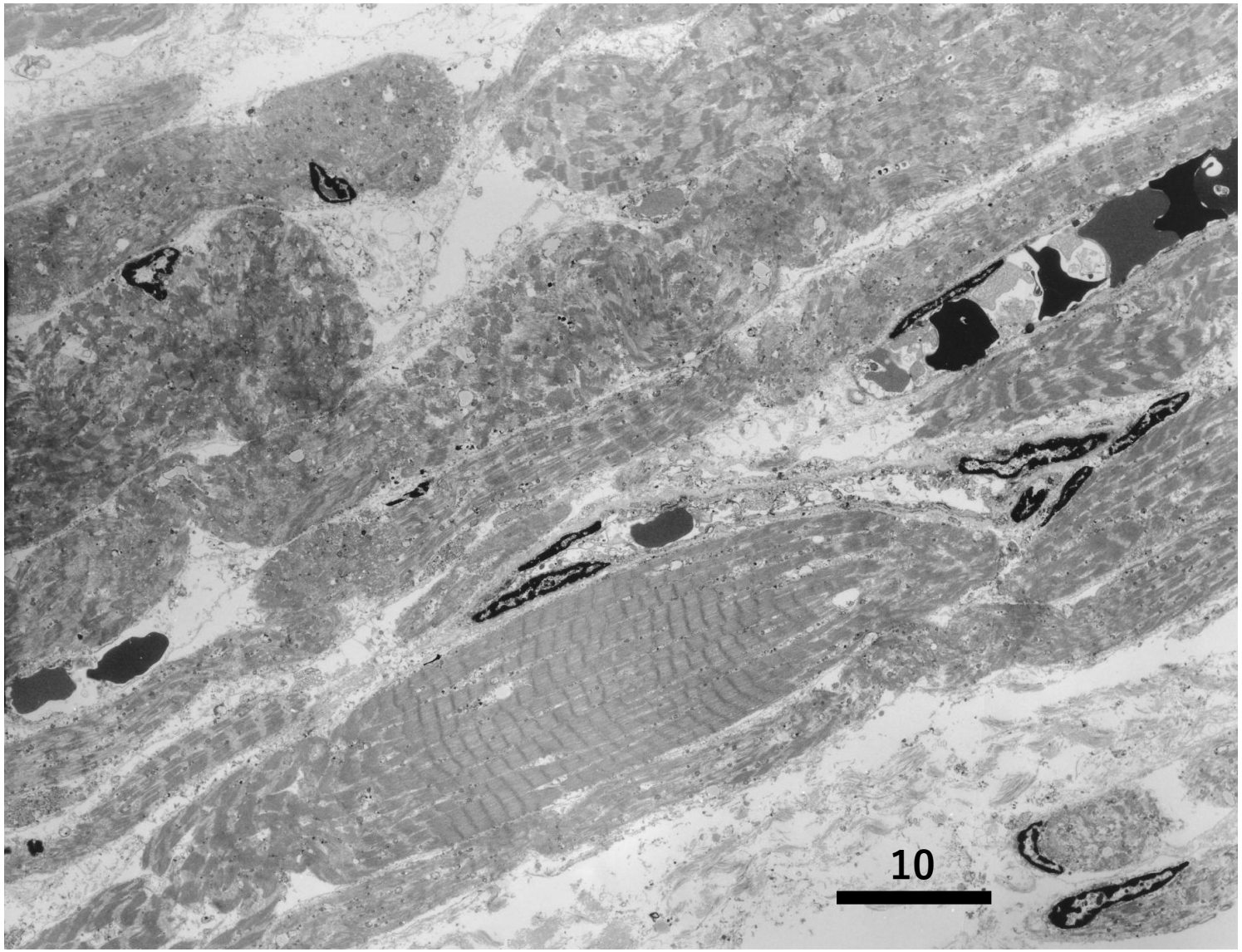
Severe lethal form of congenital myopathy with fiber-type disproportion is presented. Intrauterine fetal death occurred at the 34^{th} gestational week. At autopsy, the female baby showed general edema and retention of ascites and pleural effusion. Psoas muscle was sampled.



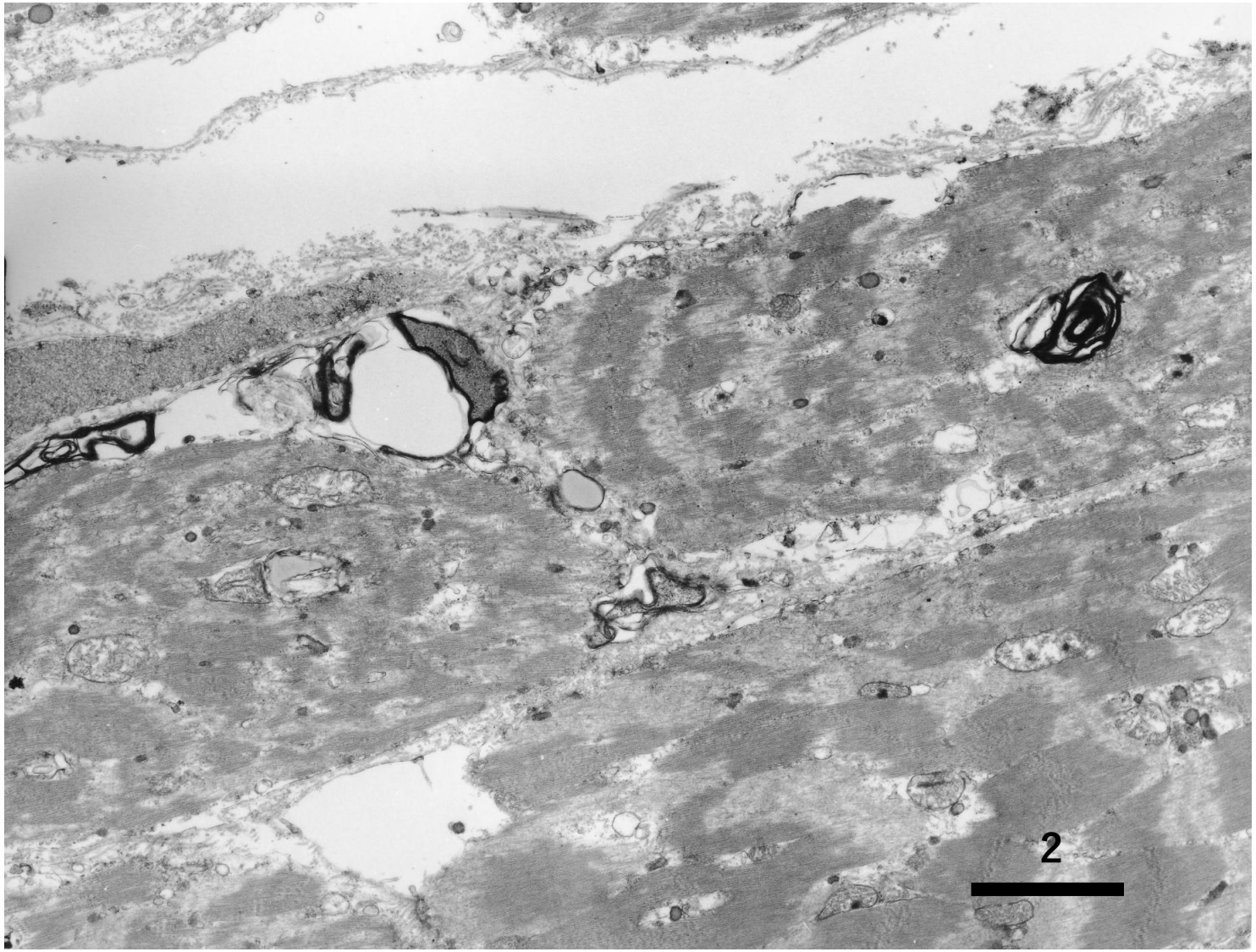
Congenital myopathy with fiber-type disproportion, severe lethal form. Intrauterine fetal death occurred at the 34th gestational week. The psoas muscle is composed of markedly thin muscle fibers. H&E-1



Congenital myopathy with fiber-type disproportion, severe lethal form. Intrauterine fetal death occurred at the 34th gestational week. The psoas muscle is composed of markedly thin muscle fibers. H&E-2



EM features of congenital myopathy with fiber-type disproportion, severe lethal form (female baby at the 34th week of gestation). Dysorganization of Z-banding is evident. EM-1

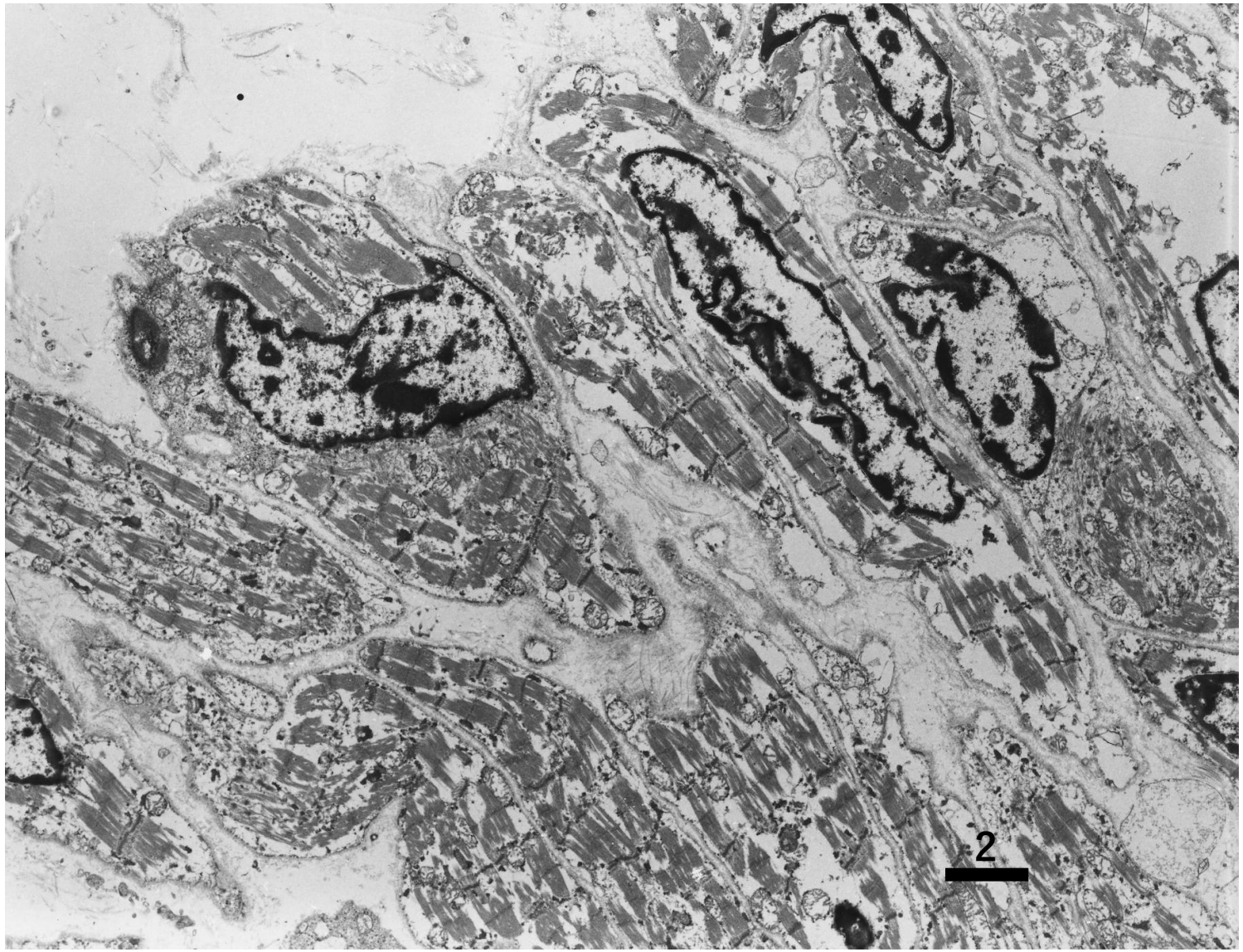


EM features of congenital myopathy with fiber-type disproportion, severe lethal form (female baby at the 34th week of gestation). Dysorganization of Z-banding is evident. EM-2

Congenital muscular dystrophy

Congenital muscular dystrophy is a muscle weakness disorder presenting early in infancy or soon after birth and gradually progressive with age. It is caused by mutation of the gene encoding dystroglycan, and the mutation is common with muscular dystrophy of Walker-Warburg type and Fukuyama type (dystroglycanopathies).

An autopsy case of 8-day-old male neonate case is presented for reference.



EM features of congenital muscular dystrophy (8-day-old male neonate). Muscle fibers are thin, but dysorganization of Z-banding is not observed. EM