



Przypadek 6/10 (7/10 ME)

Rozpoznanie: podejrzenie miopatii

Fig. 1,2,3,4. Brak istotnych zmian w obrębie włókien mięśniowych, prawidłowy układ miofibryli. W obrębie niektórych mitochondriów obserwuje się ubytek grzebieni mitochondrialnych.

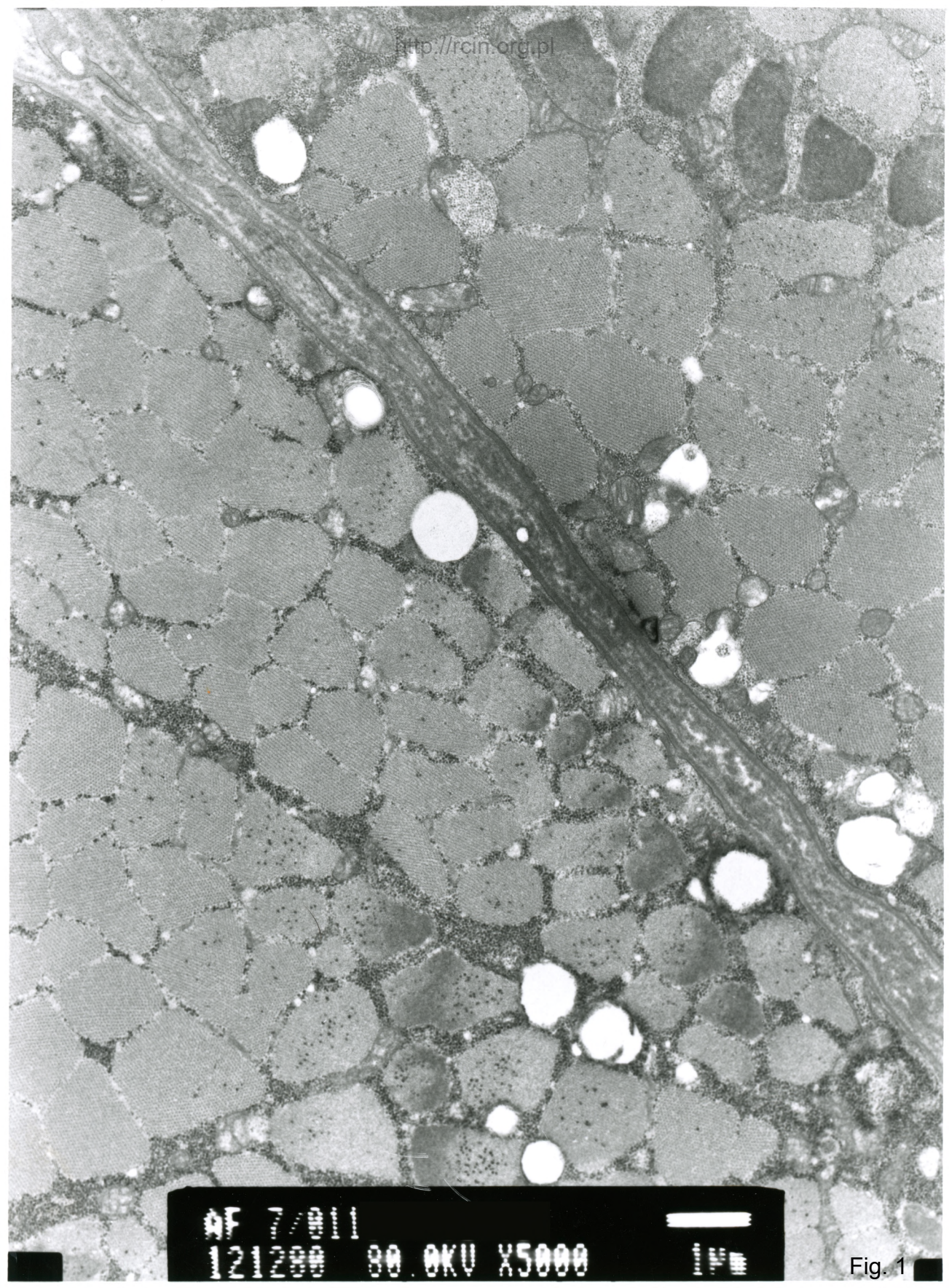
Fig. 5. Widoczne krople tłuszczu.

### Summary

A 19-year-old patient with suspected myopathy was examined. A biopsy of *quadriceps sinister* was performed.

No significant ultrastructural changes in muscle fibers were observed. Some mitochondria were characterized by loss of mitochondrial cristae (Fig. 1,2,3,4). Electronmicroscopy analysis revealed also the presence of fat droplets within the muscle fibers (Fig. 5).

A defect of mitochondrial enzymes was suspected. Biochemical examination of carnitine palmitoyltransferase was performed and it showed enzyme deficiency. The result of carnitine palmitoyltransferase level was 4,38 nM/mgB/min. (normal range 6,5-18 nM/mgB/min.).



AF 7/011  
121290 80.0KV X5000

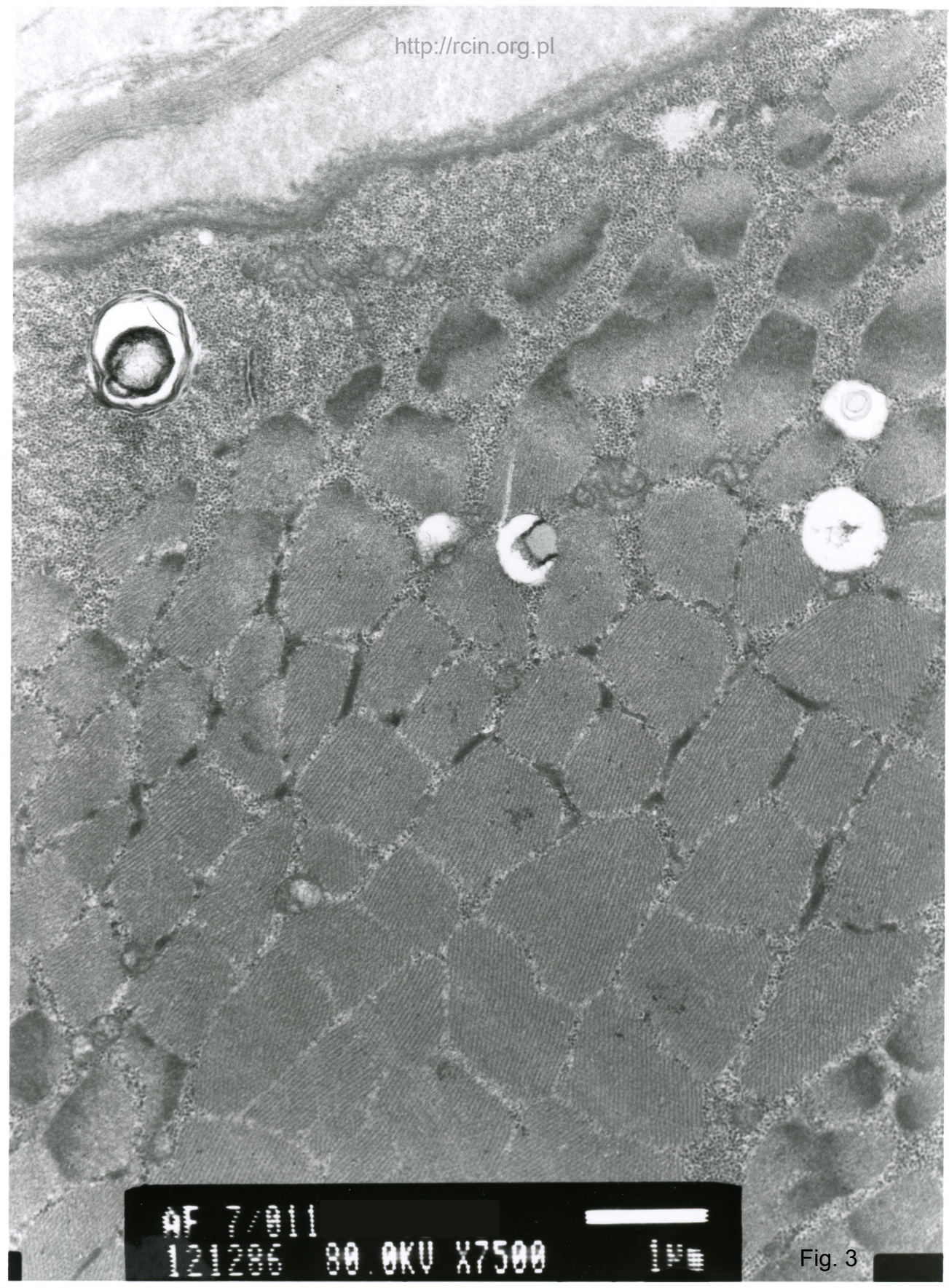


Fig. 1



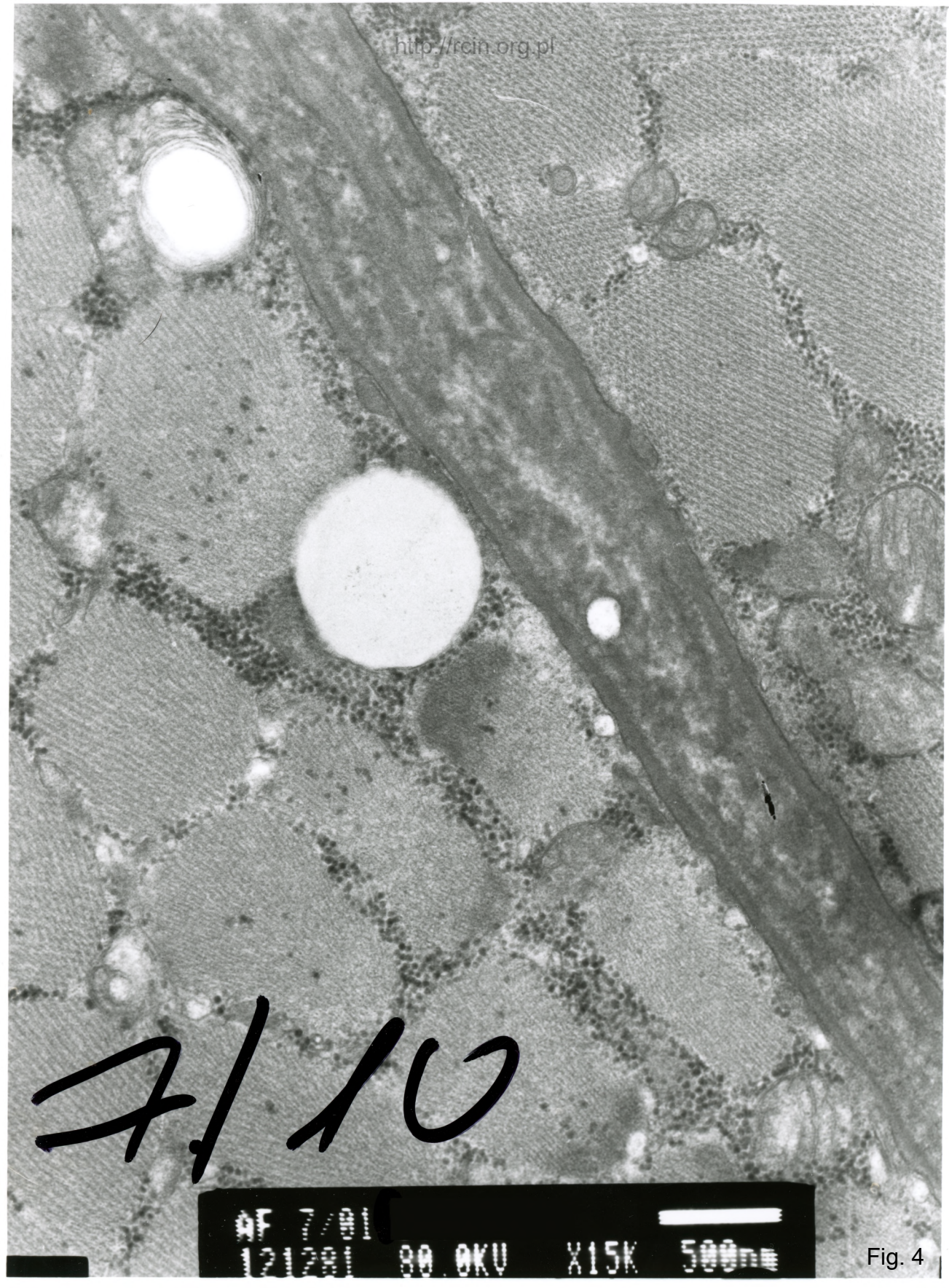
AF 7/011  
121282 80.0KV X10K 500nm

Fig. 2



AF 7/011  
121286 80.0KV X7500

Fig. 3



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AF 7/01  
121291 80.0KV X15K 500nm

Fig. 4

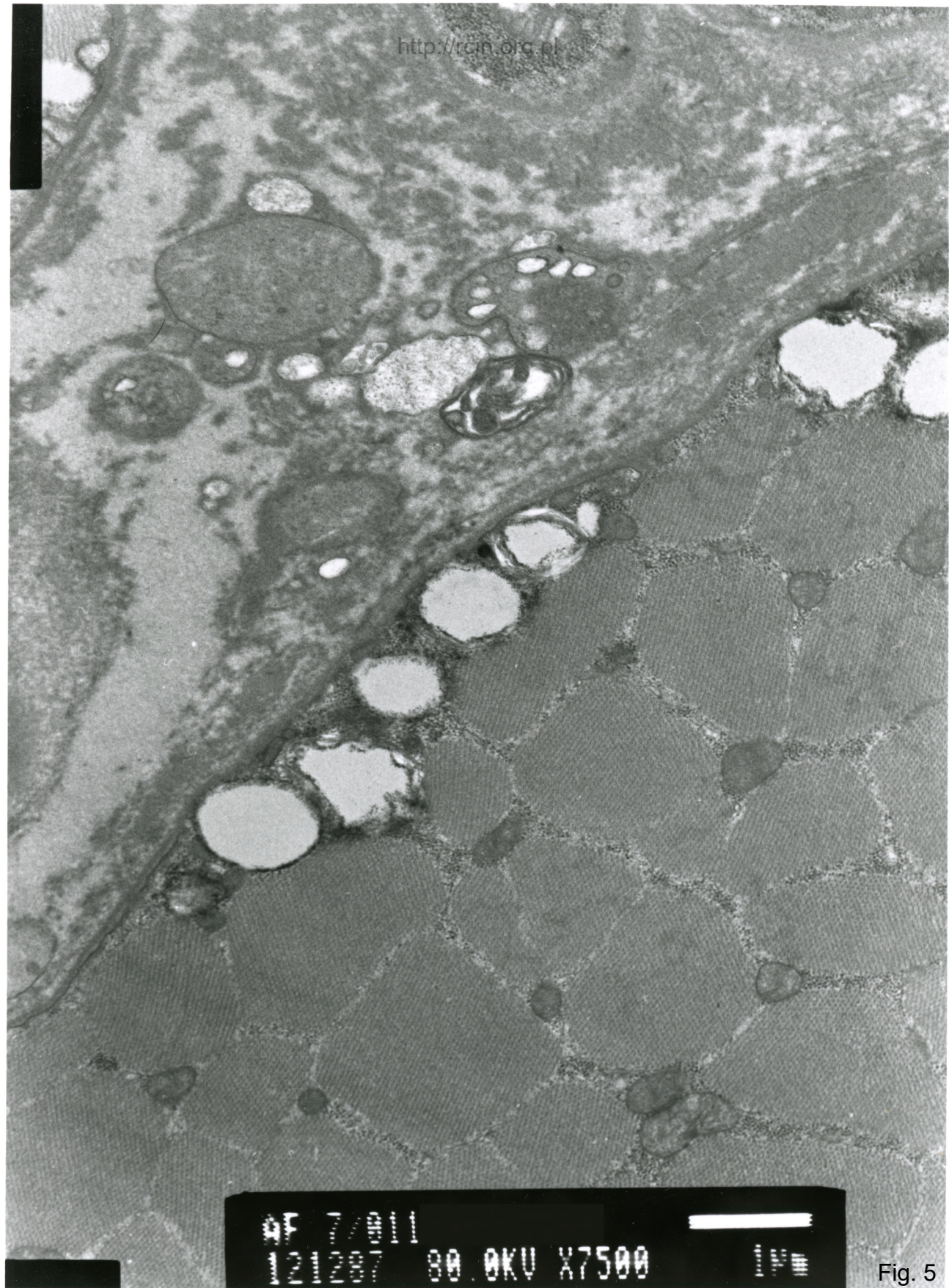


Fig. 5