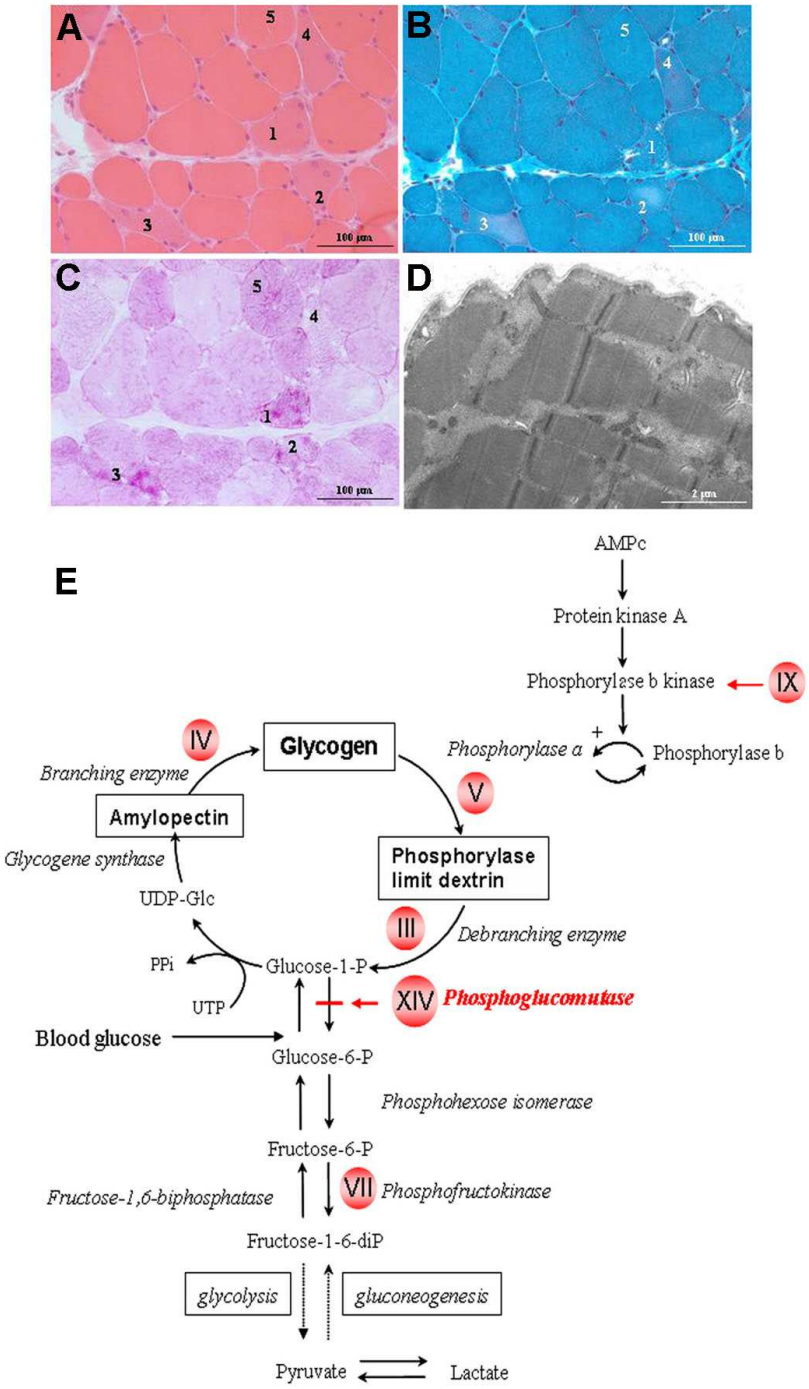
Supplementary Appendix

This appendix has been provided by the authors to give readers additional information about their work.

Supplement to: Stojkovic T, Vissing J, Petit F, et al. Muscle glycogenosis due to phosphoglucomutase 1 deficiency. N Engl J Med 2009;361:425-7.



Supplementary Figure 2: Light and electron microscopic findings in muscle biopsy (A-D). Serial transverse sections of vastus lateralis muscle are shown on Haematoxylin Eosin (A), Gomori's trichome (B) and Periodic Acid Schiff (C) stains. Numbers indicate corresponding affected fibers in the serial sections. The biopsy shows an abnormal variation in fiber diameter and many internalized nuclei. Some fibers (1-4) show a slightly basophilic cytoplasm, large vesicular nuclei, a few small vacuoles (1 and 3), and abnormal glycogen deposits. Other fibers (5) show only a slightly increased amount of glycogen. Ultrastructural data reveal accumulations of normally structured glycogen localized both intrasarcoplasmic and in the subsarcolemmal space (D). Glycogen metabolism in muscle and the various glycogen storage diseases (E), including PGM deficiency (type XIV).