

Expanded View Figures

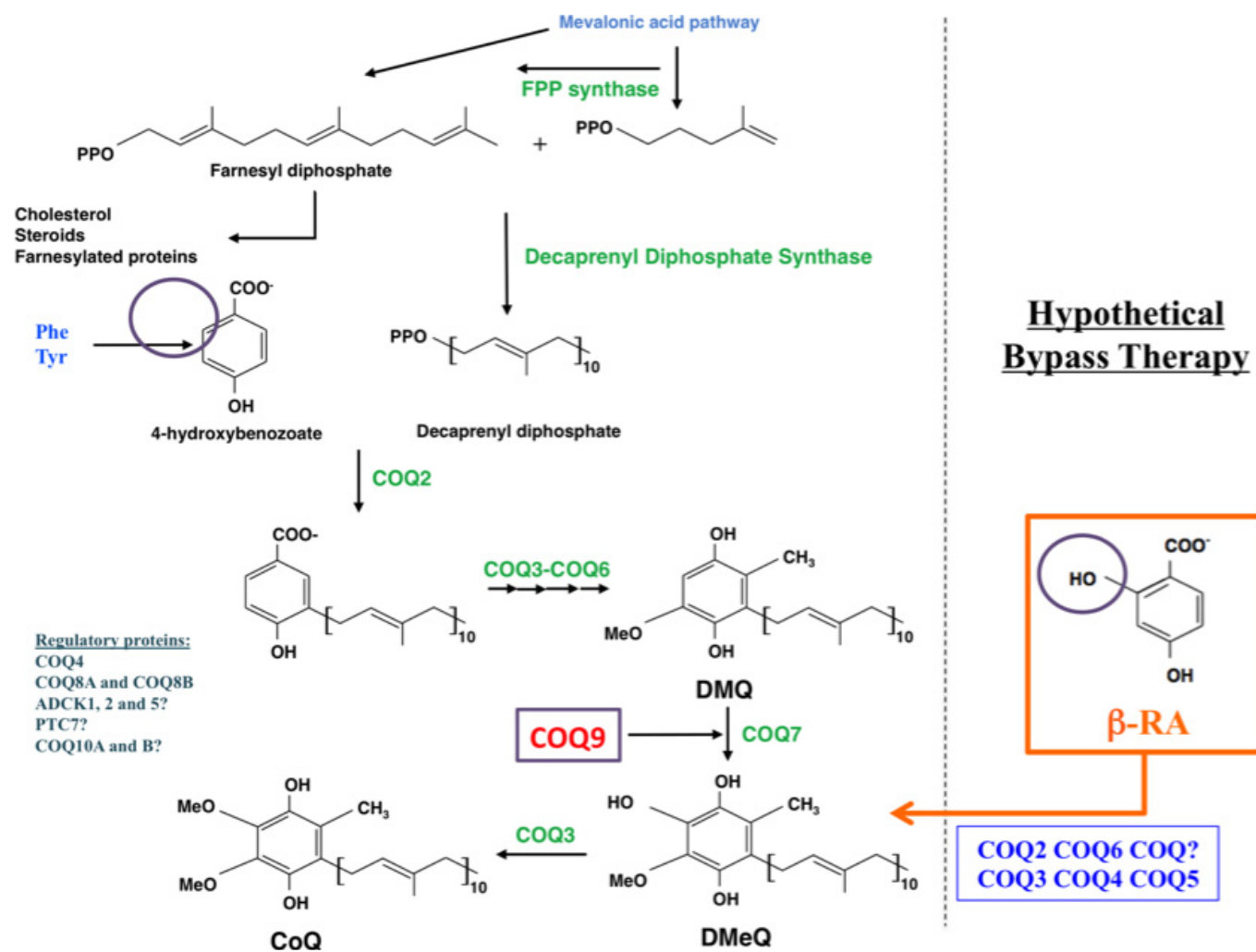


Figure EV1. CoQ biosynthetic pathway, including the bypass hypothesis for defects in COQ9 or COQ7.

4-HB = 4-hydroxybenzoic acid; DMQ = demethoxyubiquinone or 2-polyprenyl-6-methoxy-3-methyl-1,4-benzoquinone; DMeQ = 2-polyprenyl-5-hydroxy-6-methoxy-3-methyl-1,4-benzoquinone; β-RA = β-resorcylic acid. The purple circle indicates the OH group present in the β-RA and absent in the 4-HB. The hydroxylation of DMQ is normally catalyzed by COQ7, which needs COQ9 for its stability and activity. This step is compromised in patients with defects in COQ7 or COQ9. The pathway is exemplified with the production of CoQ₁₀.

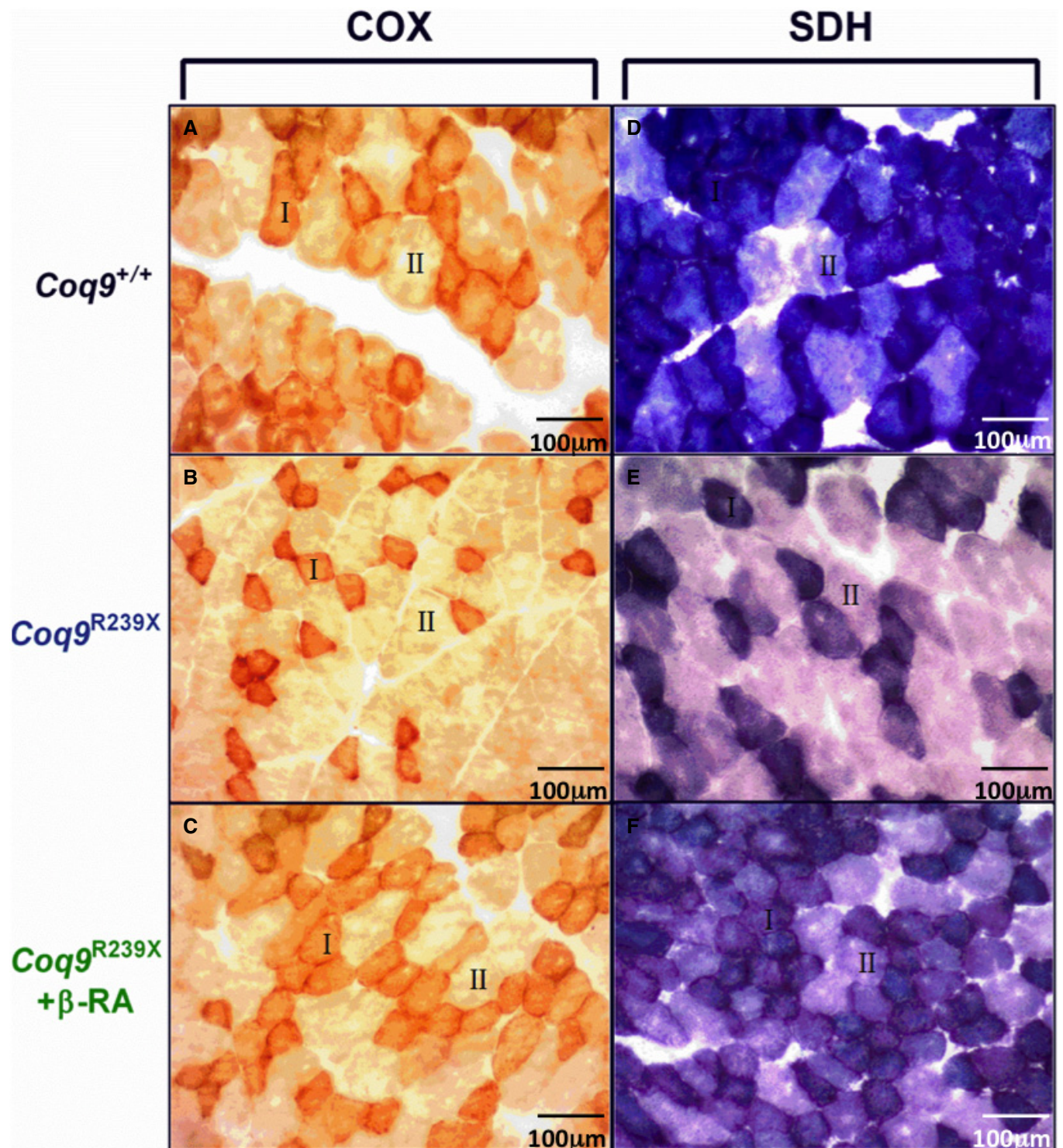


Figure EV2. COX and SDH staining in gastrocnemius.

A–C COX stains in gastrocnemius of *Coq9*^{+/+} mice (A), *Coq9*^{R239X} mice (B), and *Coq9*^{R239X} mice after β -RA treatment (C).
D–F SDH stain in gastrocnemius of *Coq9*^{+/+} mice (D), *Coq9*^{R239X} mice (E), and *Coq9*^{R239X} mice after β -RA treatment (F).

Data information: “I” indicated fiber types I and “II” indicates fiber types II.