

Publications:

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- 2) Quinzii C, Naini A, Salviati L, **Trevisson E**, Navas P, Dimauro S, Hirano M. (2006). A Mutation in Para-Hydroxybenzoate-Polyprenyl Transferase (COQ2) Causes Primary Coenzyme Q10 Deficiency. **American Journal of Human Genetics**. 78:345-9.
- 3) Martella M, Salviati L, Casarin A, **Trevisson E**, Opocher G, Polli R, Gross D, Murgia A. (2006) Molecular analysis of two uncharacterized sequence variants of the VHL gene. **Journal of Human Genetics**. 51:964-8.
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- 9) Sacconi S, Salviati L, Nishigaki Y, Walker WF, Hernandez-Rosa E, **Trevisson E**, Delplace S, Desnuelle C, Shanske S, Hirano M, Schon EA, Bonilla E, De Vivo DC, DiMauro S, Davidson MM. (2008) A functionally dominant mitochondrial DNA mutation. **Human Molecular Genetics**. 17:1814-20.
- 10) Montero R, Sánchez-Alcázar JA, Briones P, Hernández AR, Cordero MD, **Trevisson E**, Salviati L, Pineda M, García-Cazorla A, Navas P, Artuch R. (2008) Analysis of Coenzyme Q10 in muscle and fibroblasts for the diagnosis of CoQ10 deficiency syndromes. **Clinical Biochemistry**. 41:697-700.
- 11) Casarin A, Jimenez-Ortega JC, **Trevisson E**, Pertegato V, Doimo M, Ferrero-Gomez ML, Abbadi S, Artuch R, Quinzii C, Hirano M, Basso G, Ocaña CS, Navas P, Salviati L. (2008) Functional characterization of human *COQ4*, a gene required for Coenzyme Q10 biosynthesis. **Biochemical and Biophysical Research Communications**. 372:35-9.
- 12) Sacconi S, Salviati L, **Trevisson E**. (2009) Mutation analysis of COX18 in 29 patients with isolated Cytochrome *c* Oxidase deficiency. **Journal of Human Genetics**. 54:419-21.
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- 14) Casarin A, Rusalen F, Doimo M, **Trevisson E**, Carraro S, Clementi M, Tenconi R, Baraldi E, Salviati L. (2009) X-linked brachytelephalangic chondrodysplasia punctata: A simple trait that is not so simple. **American Journal of Medical Genetics Part A**. 149A:2464-2468.
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- 21) Salviati L, **Trevisson E**, Rodriguez Hernandez MA, Casarin A, Pertegato V, Doimo M, Cassina M, Agosto C, Desbats MA, Sartori G, Sacconi S, Memo L, Zuffardi O, Artuch R, Quinzii C, Dimauro S, Hirano M, Santos-Ocaña C, Navas P. (2012) Haploinsufficiency of COQ4 causes coenzyme Q10 deficiency. **J Med Genet**.49:187-91.
- 22) Casarin A, Giorgi G, Pertegato V, Siviero R, Cerqua C, Doimo M, Basso G, Sacconi S, Cassina M, Rizzuto R, Brosel S, M Davidson M, Dimauro S, Schon EA, Clementi M, **Trevisson E***, Salviati L*. (2012) Copper and

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(*=**equal contribution**)

- **CHAPTER OF BOOKS**

Trevisson, E, Salviati L, Scorrano, L. OPA1 and Its Clinical Implications. In: Encyclopedia of Life Sciences (ELS). John Wiley & Sons, Ltd: Chichester. (December 2009) DOI: 10.1002/9780470015902.a0021785.

- **MUTATION REPORTS**

Trevisson E, Salviati L, Baldoin MC, Casarin A, Basso G, Burlina A. (2008) Gene symbol: ASL. Disease: Argininosuccinate lyase deficiency. *Human Genetics.* 124:303.