# **Supplemental Data**

## Submicroscopic Duplications of the Hydroxysteroid

## Dehydrogenase HSD17B10 and the E3 Ubiquitin Ligase

#### HUWE1 Are Associated with Mental Retardation

Guy Froyen, Mark Corbett, Joke Vandewalle, Irma Jarvela, Owen Lawrence, Cliff Meldrum, Marijke Bauters, Karen Govaerts, Lucianne Vandeleur, Hilde Van Esch, Jamel Chelly, Damien Sanlaville, Hans van Bokhoven, Hans-Hilger Ropers, Frederic Laumonnier, Enzo Ranieri, Charles E. Schwartz, Fatima Abidi, Patrick S. Tarpey, P Andrew Futreal, Annabel Whibley, F. Lucy Raymond, Michael R Stratton, Jean-Pierre Fryns, Rodney Scott, Maarit Peippo, Marjatta Sipponen, Michael Partington, David Mowat, Michael Field, Anna Hackett, Peter Marynen, Gillian Turner, and Jozef Gécz

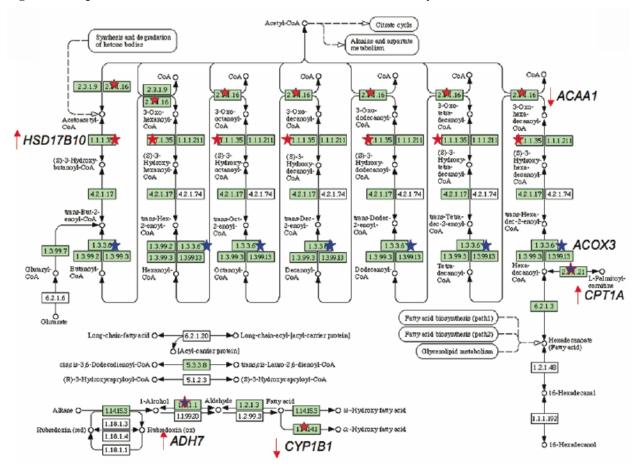
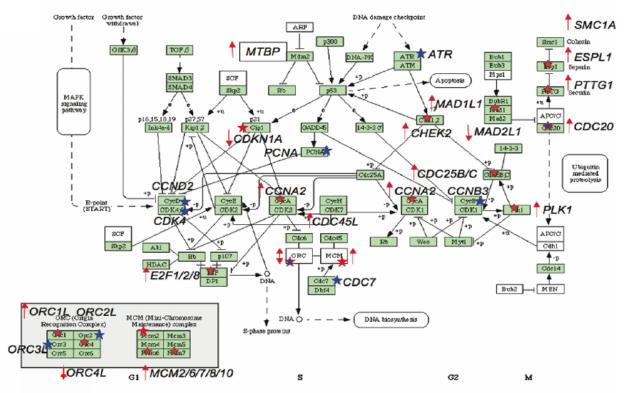


Figure S1. Duplications of *HSD17B10* Affect Cellular Metabolism Pathways

Genes that were differentially expressed (red stars), alternatively spliced (blue stars), or both (blue star with red outline) were compared to the fatty-acid metabolism pathway (hsa00071) that involve *HSD17B10*. Arrows indicate whether a gene is upregulated or downregulated.



#### Figure S2. Duplications of HUWE1 Affect Cell-Cycle Pathways

Genes that were differentially expressed (red stars), alternatively spliced (blue stars), or both (blue star with red outline) were compared to biochemical pathways that were known to involve *HUWE1*. The genes that control G1-to-S-phase transition as well as G2-to-M-phase transitions of the cell cycle (hsa04110) were affected by overexpression of *HUWE1*. Interestingly, *SMC1A*-associated genes were also affected. Genes that are not included on the KEGG pathway but may be involved are indicated on the figure enclosed in boxes. Arrows indicate whether a gene is upregulated or downregulated.