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Mammalian ultraconserved elements are strongly depleted among segmental duplications and copy number variants

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An earlier search in the human, mouse and rat genomes for sequences that are 100% conserved in orthologous segments and ≥200 bp in length identified 481 distinct sequences[1]. These human-mouse-rat sequences, which represent ultraconserved elements (UCEs), are believed to be important for functions involving DNA binding, RNA processing and the regulation of transcription and development. *In vivo* and additional computational studies of UCEs and other highly conserved sequences are consistent with these functional associations, with some observations indicating enhancer-like activity for these elements[1, 2, 3, 4, 5, 6, 7, 8, 9]. Here, we show that UCEs are significantly depleted among segmental duplications and copy number variants. Notably, of the UCEs that are found in segmental duplications or copy number variants, the majority overlap exons, indicating, along with other findings presented, that UCEs overlapping exons represent a distinct subset.

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