Whole Genome Sequence, Variant Discovery and Annotation in Mapuche-Huilliche Native South Americans

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Whole human genome sequencing initiatives help us understand population history and the basis of genetic diseases. Current data mostly focuses on Old World populations, and the information of the genomic structure of Native Americans, especially those from the Southern Cone is scant. Here we present annotation and variant discovery from high-quality complete genome sequences of a cohort of 11 Mapuche-Huilliche individuals (HUI) from Southern Chile. We found approximately 3.1 × 10 6 single nucleotide variants (SNVs) per individual and identified 403,383 (6.9%) of novel SNVs events. Analyses of large-scale genomic events detected 680 copy number variants (CNVs) and 4,514 structural variants (SVs), including 398 and 1,910 novel events, respectively. Global ancestry

composition of HUI genomes revealed that the cohort represents a sample from a marginally admixed population from the Southern Cone, whose main genetic component derives from Native American ancestors. Ad