Figure S4. Genomic region at 17q22 with suggestive evidence of association with DS-associated AVSD.
Panels A and B show the locus zoom plot and UCSC image at the corresponding genomic region. A. Locus zoom plot of the region of association at SNP with strongest signal (rs7225247). Linkage disequilibrium (LD) between this SNP (purple diamond) and nearby markers is color-coded based on the strength of LD. The left Y-axis shows the $-\log_{10}$ of the association p-value and the right Y-axis indicates the recombination rate across each region. The position on the chromosome (hg19) and the nearby genes are shown below the X-axis. B. UCSC browser image showing evidence of regulatory activity. Included are a custom track showing the location of most significant SNPs and the annotation tracks from ENCODE showing regulatory activity.