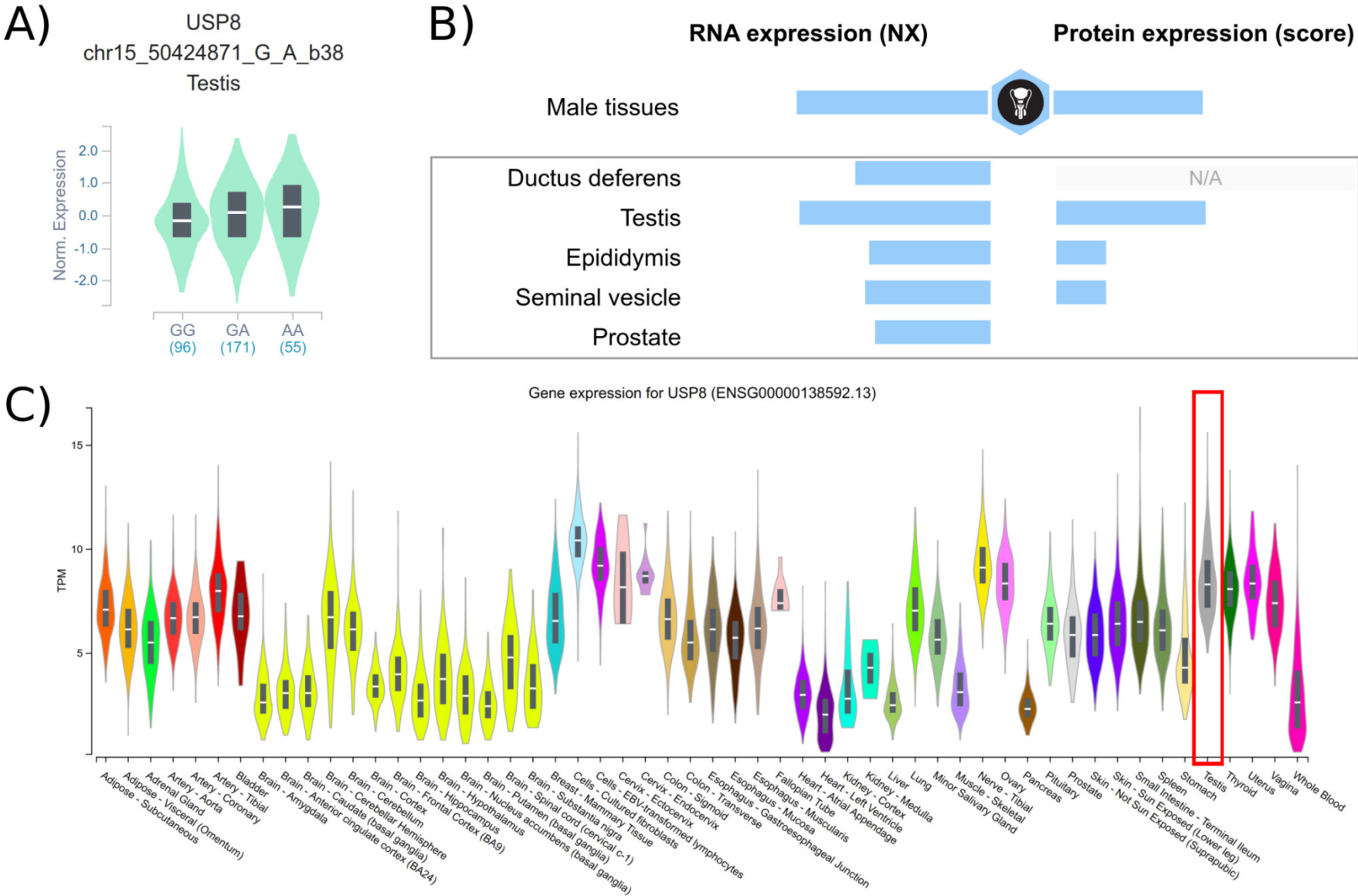
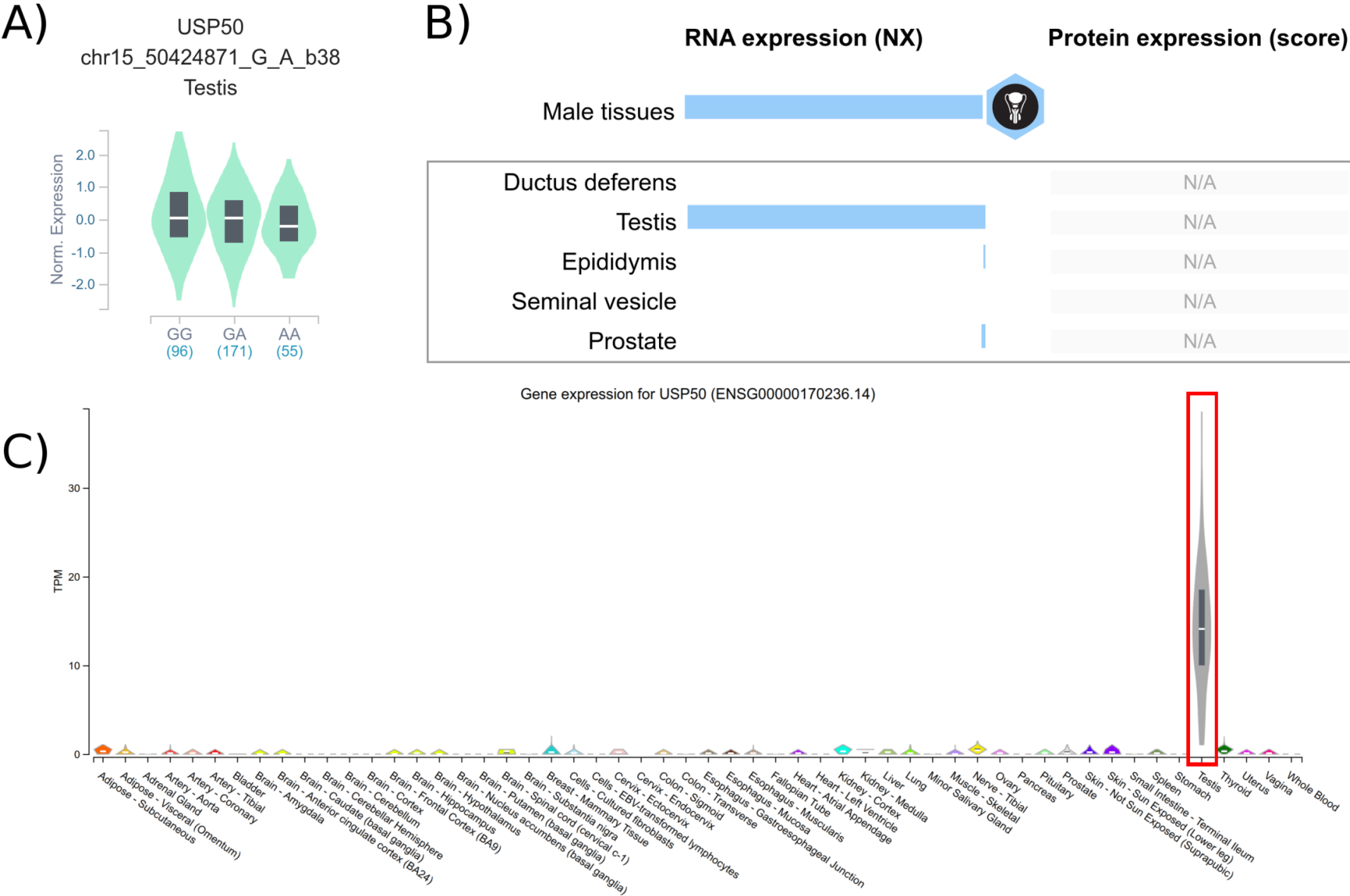


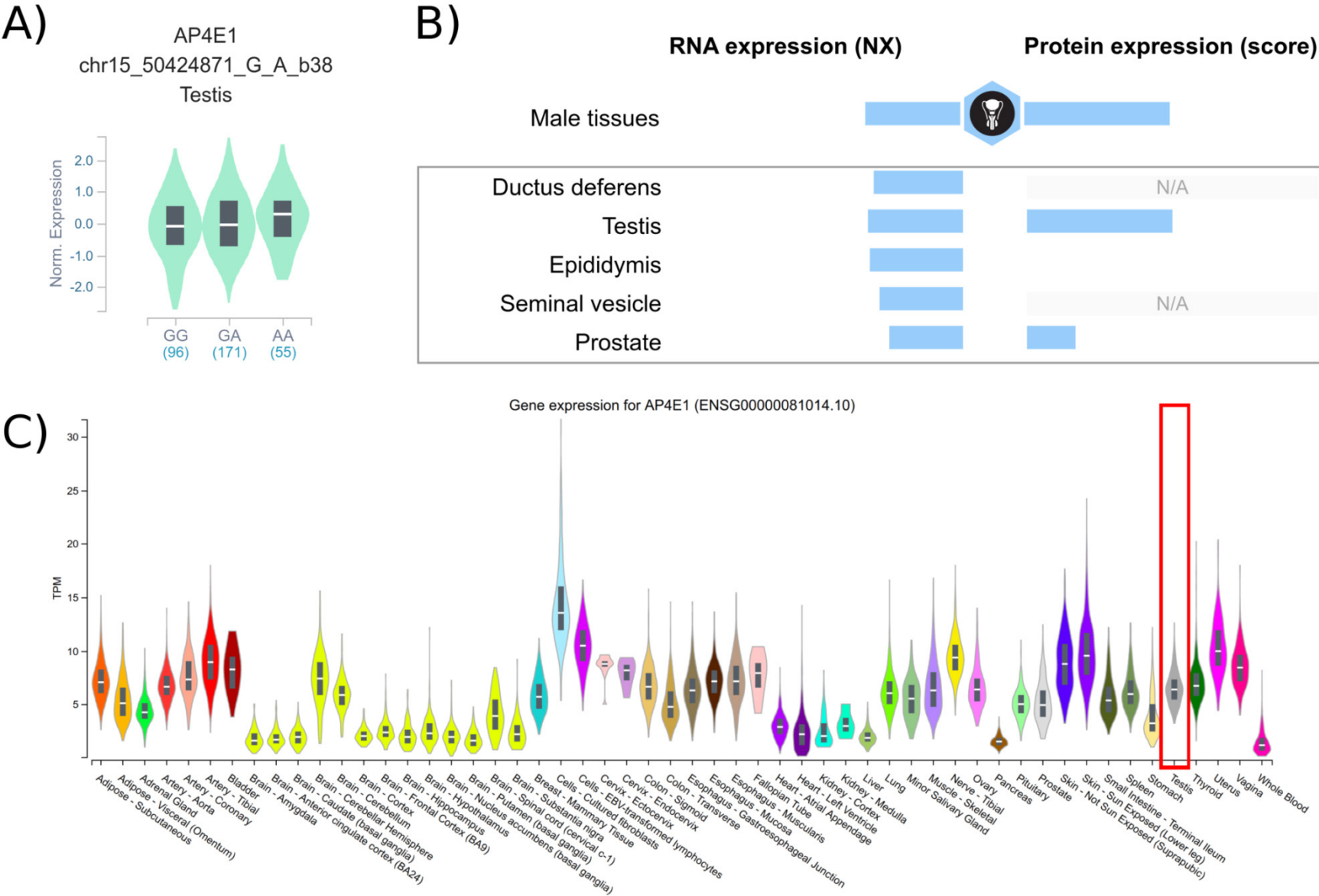
**Figure S1.** Gene expression pattern of *USP8* in human testes. **(A)** Violin plot representation of allele-specific cis-eQTL effects on *USP8* accordingly with rs7174015 genotypes in human testicular tissue of the Genotype-Tissue Expression (GTEx, analysis release v8 and human genome build 38) database. The G and A alleles indicate the reference and alternative allele types, respectively. **(B)** RNA and protein normalized expression of *USP8* in male reproductive tissues according to the Human Protein Atlas database. **(C)** *USP8* gene expression in different human tissues of the GTEx database (analysis release v8). Testis expression is highlighted with red boxes. Teal regions indicate the density distribution of the samples and the white line in the box plots the median value of the expression.



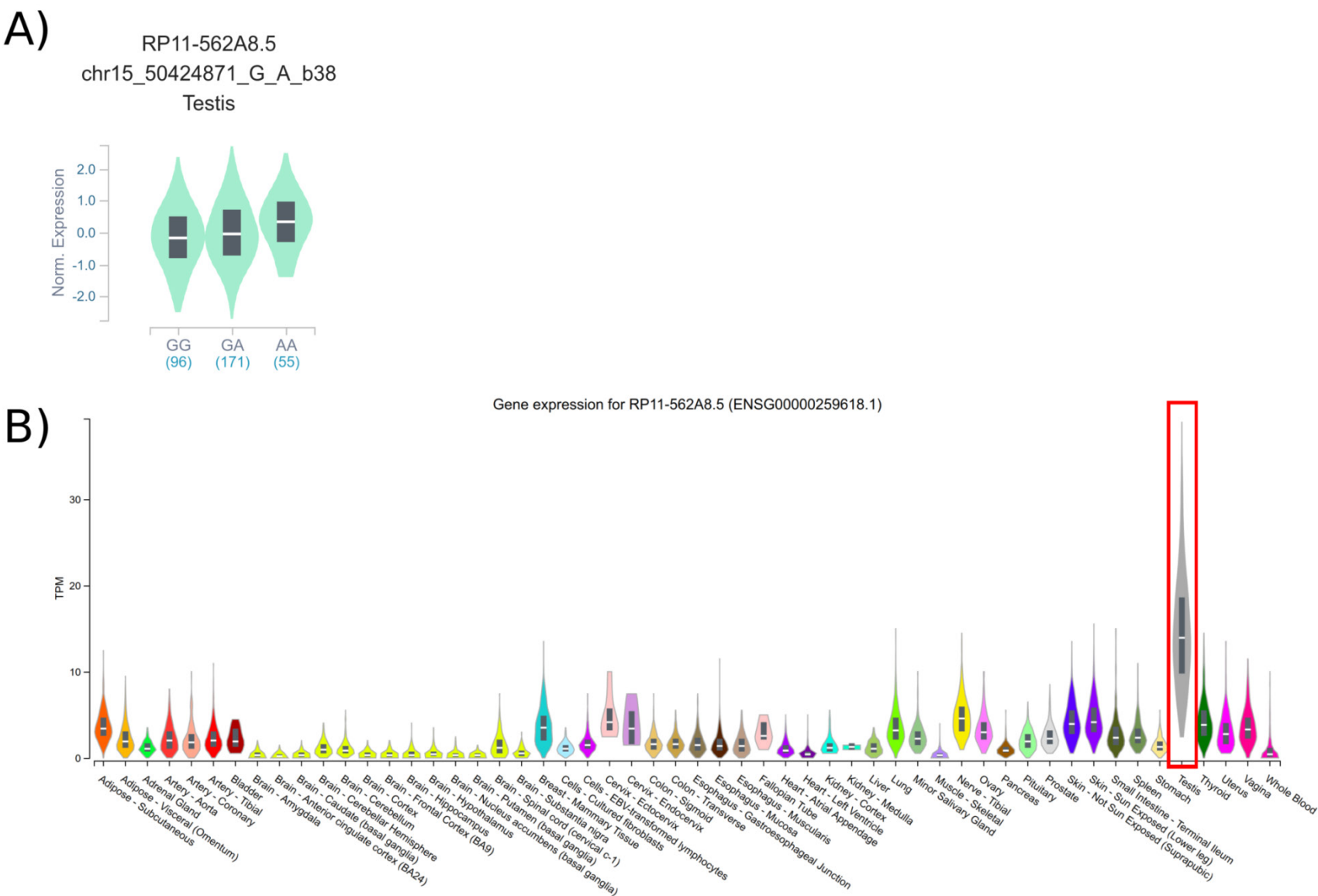
**Figure S2.** Gene expression pattern of *USP50* in human testes. **(A)** Violin plot representation of allele-specific cis-eQTL effects on *USP50* accordingly with rs7174015 genotypes in human testicular tissue of the Genotype-Tissue Expression (GTEx, analysis release v8 and human genome build 38) database. The G and A alleles indicate the reference and alternative allele types, respectively. **(B)** RNA normalized expression of *USP50* in male reproductive tissues according to the Human Protein Atlas database. **(C)** *USP50* gene expression in different human tissues of the GTEx database (analysis release v8). Testis expression is highlighted with red boxes. Teal regions indicate the density distribution of the samples and the white line in the box plots the median value of the expression.



**Figure S3.** Gene expression pattern of *AP4E1* in human testes. **(A)** Violin plot representation of allele-specific cis-eQTL effects on *AP4E1* accordingly with rs7174015 genotypes in human testicular tissue of the Genotype-Tissue Expression (GTEx, analysis release v8 and human genome build 38) database. The G and A alleles indicate the reference and alternative allele types, respectively. **(B)** RNA and protein normalized expression of *AP4E1* in male reproductive tissues according to the Human Protein Atlas database. **(C)** *AP4E1* gene expression in different human tissues of the GTEx database (analysis release v8). Testis expression is highlighted with red boxes. Teal regions indicate the density distribution of the samples and the white line in the box plots the median value of the expression.



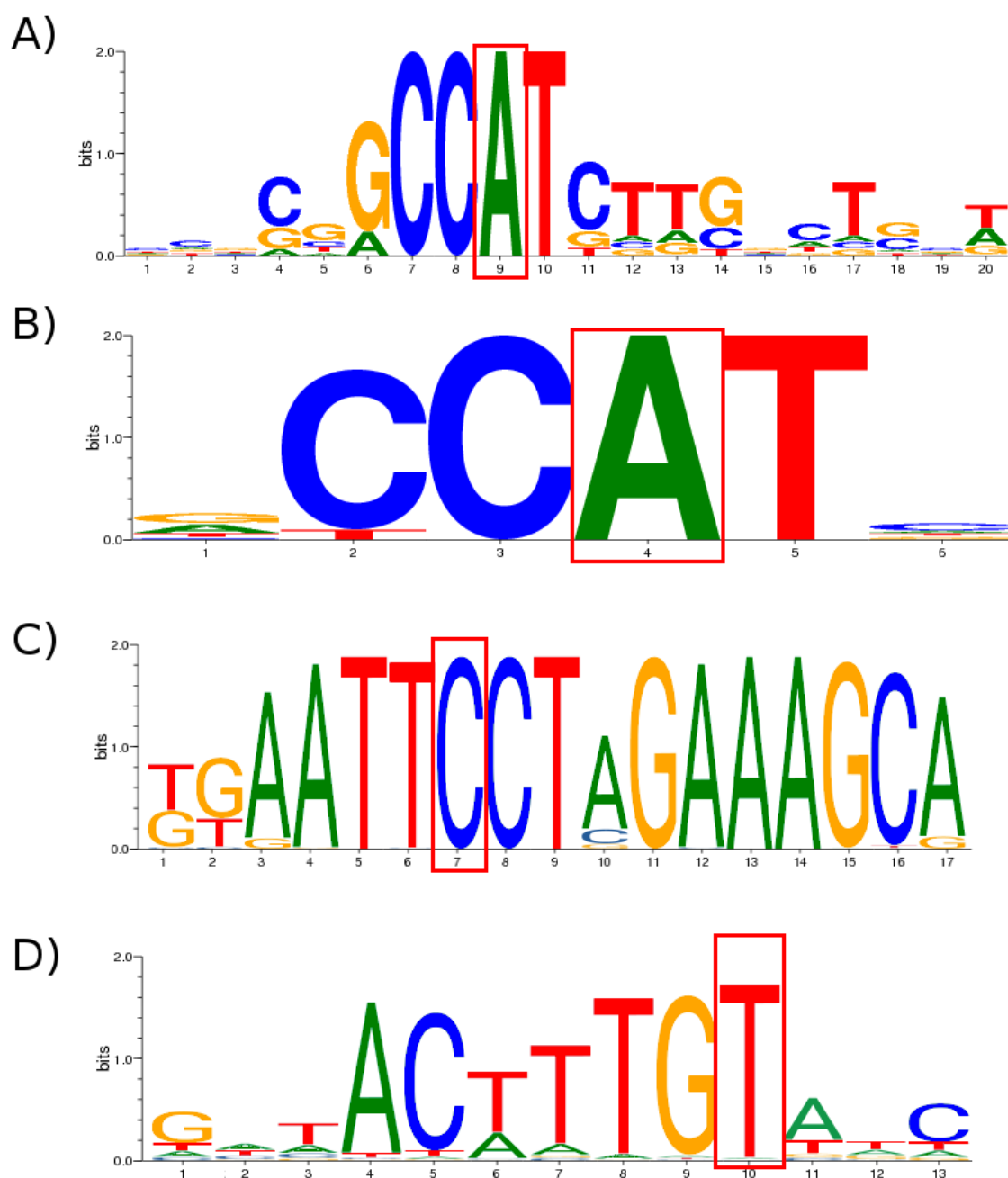
**Figure S4.** Gene expression pattern of *RP11-562A8.5* in human testes. **(A)** Violin plot representation of allele-specific cis-eQTL effects on *RP11-562A8.5* accordingly with rs7174015 genotypes in human testicular tissue of the Genotype-Tissue Expression (GTEx, analysis release v8 and human genome build 38) database. The G and A alleles indicate the reference and alternative allele types, respectively. **(B)** *RP11-562A8.5* gene expression in different human tissues of the GTEx database (analysis release v8). Testis expression is highlighted with red boxes. Teal regions indicate the density distribution of the samples and the white line in the box plots the median value of the expression.



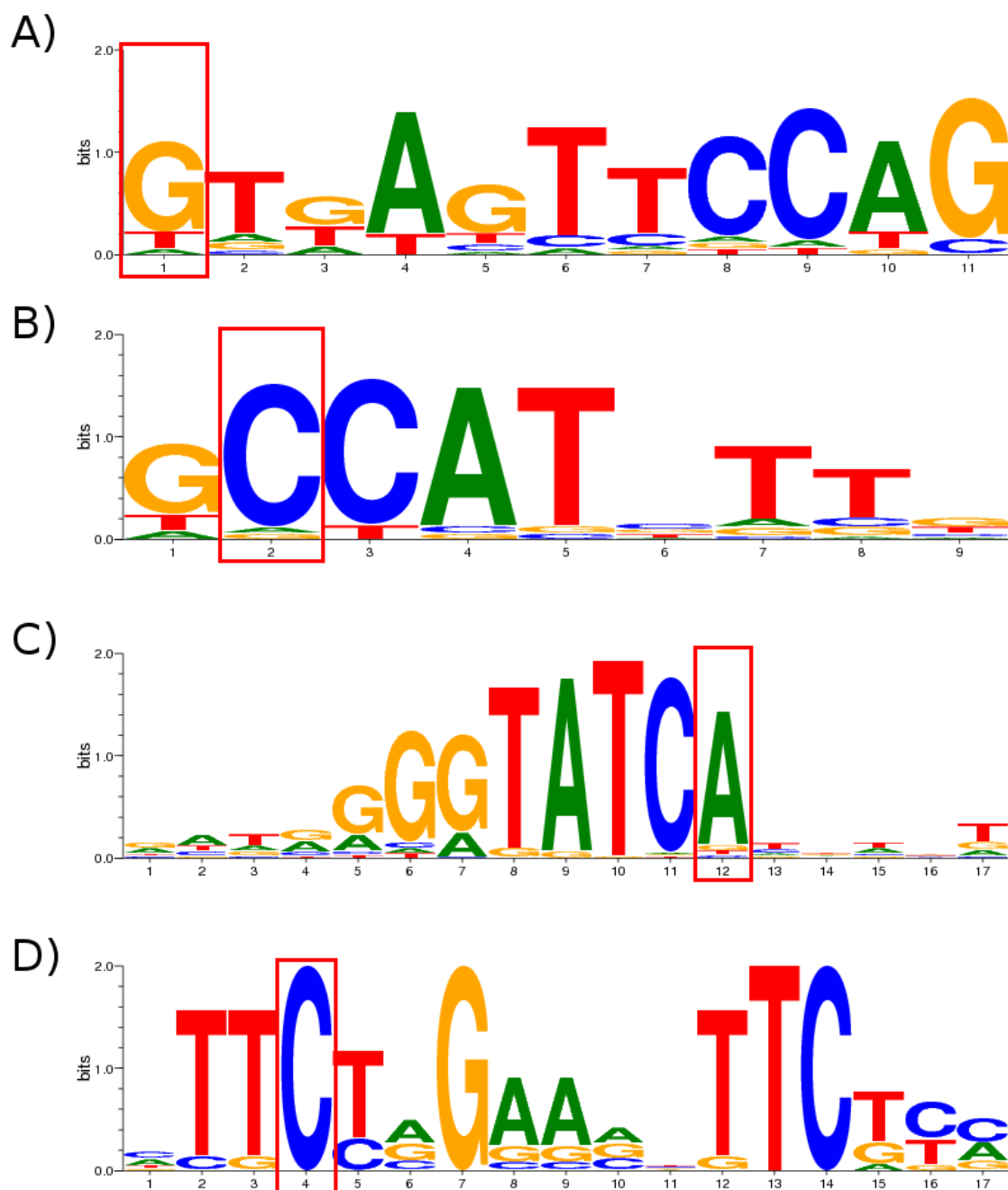




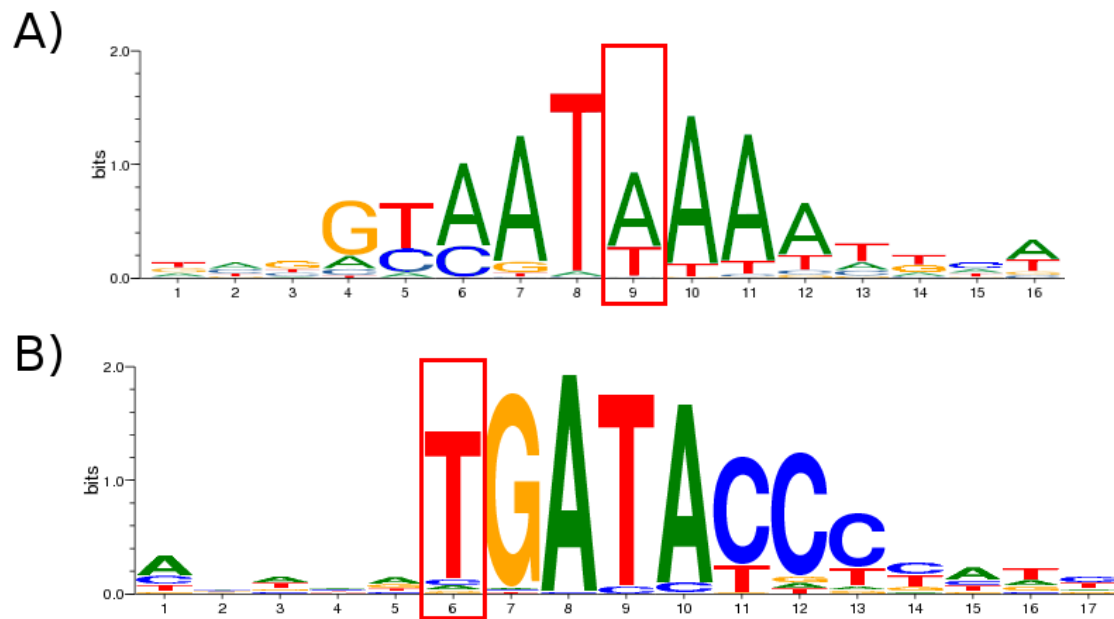
**Figure S6.** Sequence logos of Position Weight Matrices (PWM) for transcription factor binding sites strongly altered by *TUSC1*-rs10966811 proxies. The position of each proxy is highlighted in red for the following pairs of polymorphism/motif changed: **(A)** rs10966811/YY1\_known2, **(B)** rs10966811/YY1\_known6, **(C)** rs10966813/Bcl6b, **(D)** rs11789162/DMRT1.



**Figure S7.** Sequence logos of Position Weight Matrices (PWM) for transcription factor binding sites strongly altered by *USP8*-rs7174015 proxies. The position of each proxy is highlighted in red for the following pairs of polymorphism/motif changed: (A) rs12593481/Pax-5\_known3, (B) rs12593481/YY1\_known4, (C) rs28582911/SIX5\_known2, (D) rs3098171/Hsf\_known3.



**Figure S8.** Sequence logos of Position Weight Matrices (PWM) for transcription factor binding sites strongly altered by *EPSTI1*-rs12870438 proxies. The position of each proxy is highlighted in red for the following pairs of polymorphism/motif changed: (A) rs71099806/HOXA10, (B) rs9594826/SIX5\_known2.





## **Supplementary note.**

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