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Supplemental Data

A Focal Epilepsy and Intellectual Disability Syndrome Is Due to a Mutation in *TBC1D24*

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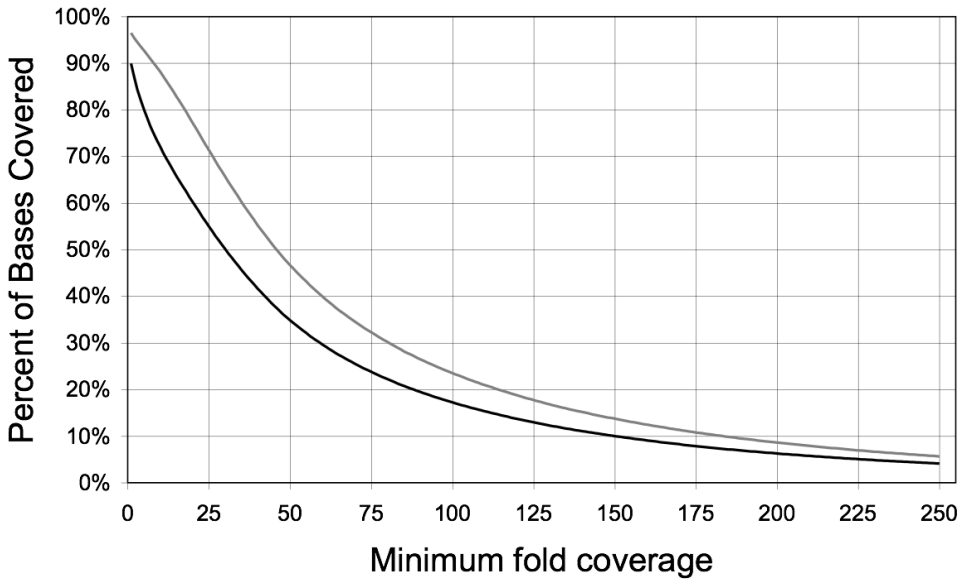

Figure S1. A Single Lane of Illumina Sequence Provides Sufficient Coverage for Identification of Sequence Variants
The percent of the total sequence in the linked interval (black line) or targeted on the array (grey line) relative to the level sequence coverage

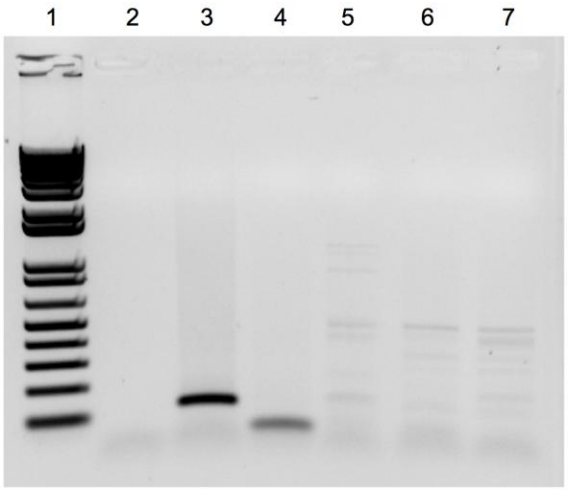

Figure S2. PRSS41 Is Not Expressed in the Human Brain
Primers specific to PRSS41 were used to amplify the product of the PRSS41 gene from cDNA derived from human testis, cortex, frontal lobe, occipital lobe and hippocampus (lanes 3 - 7 respectively). The expected product of 133bp is only visible in testis. Lane 2 shows lack of amplification from a no template control reaction, lane 1 shows product sizes indicated by the 1kb+ marker (Invitrogen).

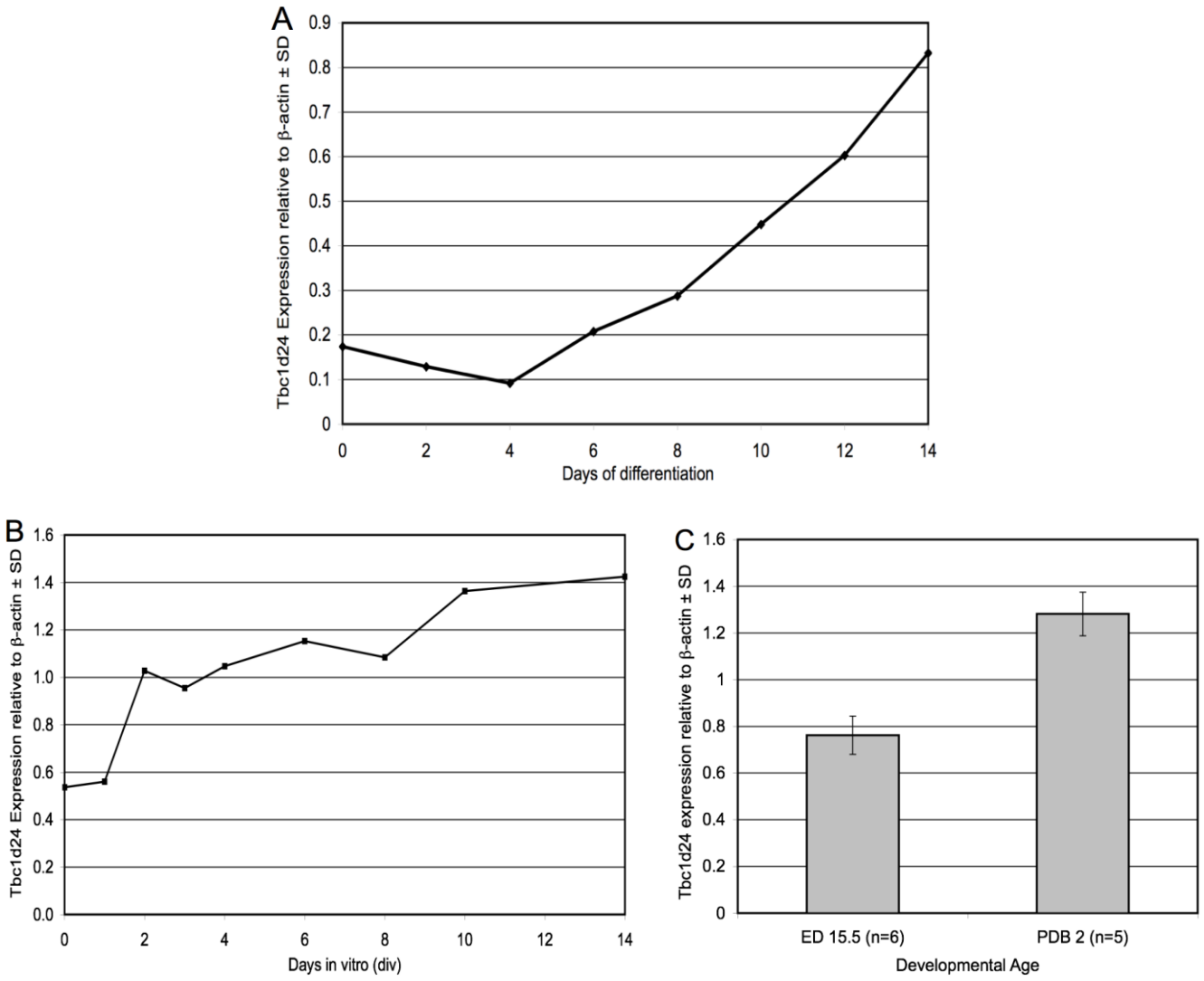

Figure S3. TBC1D24 Increases in Expression as Cells of Neuronal Origin Approach Terminal Differentiation

Expression of mouse *Tbc1d24* (primer bank ID 26350797a2) normalised to *Actb* quantified using the relative standard curve qPCR method as described previously [21] in cDNA derived from total RNA extracts from: **(A)** Mouse embryonic stem cells (ESC) directed to differentiate into cells of the neuronal lineage [22] ; **(B)** mouse embryonic day (ED) 18.5 primary hippocampal neurons cultured at 0, 1, 2, 3, 4, 6, 8, 10 and 14 days *in vitro*; and **(C)** mouse whole brain at ED15.5 and post birth day (PBD) 2. Error bars indicate standard deviations between individuals.

Table S1. Sequences Tiled on the Capture Array but Subsequently Not Assembled Are Enriched in GC Content and Repeated Sequences

	Bases Tiled but Not Covered						Bases Tiled					
	Repeats		Non-Repeats		Total		Repeats		Non-Repeats		Total	
Base	**Count**	**%**	**Count**	**%**	**Count**	**%**	**Count**	**%**	**Count**	**%**	**Count**	**%**
A	2952	13.50	10019	16.07	12971	15.40	101445	25.27	383911	19.96	485356	20.87
C	8062	36.86	21115	33.87	29177	34.65[b]	97644	24.33	570505	29.65	668149	28.73[b]
G	7701	35.21	20962	33.63	28663	34.04[b]	98540	24.55	575805	29.93	674345	29.00[b]
T	3155	14.43	10244	16.43	13399	15.91	103780	25.85	393653	20.46	497433	21.39
Total	21870	25.97[a]	62340	74.03	84210		401409	17.26[a]	1923874	82.74	2325283	

[a] Statistically significant enrichment of repeated sequences (as defined by repeat masker) compared to that expected from the total tiled content $p < 0.05$ by χ^2 analysis.
[b] Statistically significant enrichment of the combined G/C content not covered compared to that expected from the total tiled content, $p < 0.05$ by χ^2 analysis.

Table S2. Summary of All Unique Homozygous SNPs
(See accompanying Excel spreadsheet)

Supplemental References

21. Tarpey, P.S., Raymond, F.L., Nguyen, L.S., Rodriguez, J., Hackett, A., Vandeleur, L., Smith, R., Shoubridge, C., Edkins, S., Stevens, C. *et al.* (2007). Mutations in UPF3B, a member of the nonsense-mediated mRNA decay complex, cause syndromic and nonsyndromic mental retardation. Nat. Genet 39, 1127-1133
22. Ying, Q., Stavridis, M., Griffiths, D., Li, M. and Smith, A. (2003). Conversion of embryonic stem cells into neuroectodermal precursors in adherent monoculture. Nat. Biotechnol 21, 183-186.