**Supplementary Information**

**Germline and somatic mutations in *STXBP1* with diverse neurodevelopmental phenotypes**

Mohammed Uddin1,\*, Marc Woodbury-Smith1,3, Ada Chan4,5, Ledia Brunga4,Sylvia Lamoureux3,4, Giovanna Pellecchia3,4, Ryan KC Yuen3,4, Muhammad Faheem3,4,Dimitri J. Stavropoulos6, James Drake7, Cecil D Hahn8,9, Cynthia Hawkins9, Adam Shlien4, Christian R Marshall3, Lesley A Turner10, Berge Minassian4,7, Stephen W. Scherer3,4,5,11 , Cyrus Boelman12,\*

1) Mohammed Bin Rashid University of Medicine and Health Sciences, Dubai, UAE.

2) Department of Psychiatry and Behavioural Neurosciences, McMaster University, Hamilton, Ontario, Canada;

3) The Centre for Applied Genomics, The Hospital for Sick Children, Toronto, Ontario, Canada;  
4) Program in Genetics and Genome Biology (GGB), The Hospital for Sick Children, Toronto, Ontario, Canada;

5) Department of Molecular Genetics, University of Toronto, Toronto, Ontario, Canada  
6) Genome Diagnostics, Paediatric Laboratory Medicine, The Hospital for Sick Children, Toronto, Ontario, Canada;  
7) Division of Neurosurgery, The Hospital for Sick Children, Toronto, Ontario, Canada;  
8) Division of Neurology, The Hospital for Sick Children, Toronto, Ontario, Canada;  
9) Department of Paediatrics, University of Toronto, Toronto, Ontario, Canada;

10) Discipline of Genetics, Faculty of Medicine, Memorial University of Newfoundland, St. John’s, Newfoundland, Canada;  
11) McLaughlin Centre, University of Toronto, Toronto, Ontario, Canada.

12) Division of Neurology, BC Children’s Hospital, Vancouver, BC, Canada;

\*Cyrus Boelman, Division of Neurology, BC Children’s Hospital, Vancouver, BC Canada,([cyrus.boelman@cw.bc.ca](mailto:cyrus.boelman@cw.bc.ca)); Mohammed Uddin, College of Medicine, Mohammed Bin Rashid University of Medicine and Health Sciences, Dubai, UAE ([mohammed.uddin@mbru.ac.ae](mailto:mohammed.uddin@mbru.ac.ae))

**Supplementary Table e-2.** Epilepsy encephalopathy cohort analyzed to quantify the rate of *de novo* mutations from published literatures[1-33](#_ENREF_1).

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| --- | --- | --- | --- | --- |
| **Study Cohort Size** | **PMID** | **Number of *de novo* within *STXBP1*** | **Number of *de novo* mutationwithin *STXBP1*** | **% of de novo in a cohort** |
| 45 | 23020937 | 3 | 3 | 6.7 |
| 29 | 21762454 | 3 | 3 | 10.3 |
| 106 | 20876469 | 6 | 5 | 4.7 |
| 95 | 21376300 | 2 | 2 | 2.1 |
| 50 | 21364700 | 1 | 1 | 2.0 |
| 70 | 24623842 | 3 | 3 | 4.3 |
| 284 | 26514728 | 24 | 20 | 7.0 |
| 68 | 23662938 | 3 | 3 | 4.4 |
| 44 | 24781210 | 1 | 1 | 2.3 |
| 33 | 22612257 | 1 | 0 | 0.0 |
| 52 | 21770924 | 5 | 5 | 9.6 |
| 86 | 21204804 | 2 | 2 | 2.3 |
| 73 | 25497044 | 3 | 3 | 4.1 |
| 110 | 25818041 | 2 | 2 | 1.8 |
| 28 | 22211739 | 2 | 2 | 7.1 |
| 83 | 20887364 | 9 | 9 | 10.8 |
| 160 | 23409955 | 6 | 6 | 3.8 |
| 1131 | 26795593 | 3 | 3 | 0.3 |
| 22 | 27171548 | 1 | 1 | 4.5 |

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