Identification of potential pathogenic genes causing occipitalization of the atlas

Shanhang Jia, M.D.1,2, Wanru Duan, M.D.1,2, Zong Xin, M.D.1, Boyan Zhang, BMed.1, Qiang Jian, BMed.1, Chen Wei, Ph.D.,3 , Zan Chen, M.D. Ph.D. 1,2

Co-first authors Shanhang Jia, M.D. 1,2, Wanru Duan, M.D.1,2,

Corresponding authors Chen Wei, Ph.D.,3, Zan Chen, M.D. Ph.D. 1,2

1. Department of Neurosurgery, Xuanwu Hospital, Capital Medical Universiy. Beijing, China

2. Lab of Spinal Cord Injury and Functional Reconstruction, China International Neuroscience Institute (CHINA-INI)

3. Beijing Advanced Innovation Center for Biomedical Engineering, School of Biological Science and Medical Engineering, Beihang University, Beijing 100083, China

**Funded by**

1. Beijing Natural Science Foundation Grant, 7172091, Zan Chen

2. Beijing Municipal Administration of Hospitals Yang-Fan Grant, XMLX202138, Zan Chen

3. AMS/PUMC Research Project #201920200501, Human Brain Tissue Bank Platform for Neurological Diseases, Zan Chen, Wanru Duan

4. Beijing Health Commission Independent Innovation Fund (2018-2-2014), Zan Chen

**Email**

[shanhangjia@126.com](mailto:shanhangjia@126.com)

[duanwanru@xwhosp.org](mailto:duanwanru@xwhosp.org),

rediscovered@163.com

[zhangboyan1997@foxmail.com](mailto:zhangboyan1997@foxmail.com),

[88316314@qq.com](mailto:88316314@qq.com)

[chenw123@buaa.edu.cn](mailto:chenw123@buaa.edu.cn);

chenzan66@163.com,

**Supplementary Material**

This supplemental file was intended for publication as a data supplement. The file includes 1 tables cited in the manuscript.

The contents include the following:

**Table S1. List of candidate genes associated with vertebral segmentation defects as well as related diseases**

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Gene** |  |  |  |  |
| A2ML1 | DNAAF1 | INA | NPHP3 | SMAD6 |
| ABHD14A-ACY1 | DNMT3A | ITPR2 | NSD1 | SMARCA2 |
| AGAP1 | DOCK8 | JMJD1C | NT5E | SMARCAD1 |
| ALDH2 | EBP | KAT6B | NTNG1 | SMC2 |
| ALMS1 | EDNRA | KCNQ1 | OTOA | SMG7 |
| ALPL | EFTUD2 | KIF14 | PABPC1 | SMOC1 |
| AMER1 | EHMT1 | KIF22 | PAPPA | SOHLH1 |
| ANKH | EIF4G1 | KIFBP | PARD3B | SON |
| ANLN | ELN | KLHDC10 | PCSK5 | SOS2 |
| ARID3B | ELP1 | KMT2A | PCYT1A | SRC |
| ARVCF | ENG | KMT2C | PDE11A | STAT5A |
| ASPM | ENPP1 | LAMA2 | PEX5 | STIL |
| ATP7B | EPHB2 | LAMA5 | PHGDH | SZT2 |
| ATP8B3 | EXT2 | LAMC3 | PLOD3 | TBX2 |
| ATPAF2 | EZH2 | LETM1 | PMFBP1 | TBXAS1 |
| ATR | F12 | LIMCH1 | POLG | TCF12 |
| BBS9 | FAN1 | LPO | POR | TCIRG1 |
| BDNF | FANCA | LRP5 | PRKRA | TCTN2 |
| BMP2 | FANCB | LRRK1 | PRSS12 | TEX14 |
| BMS1 | FANCD2 | LZTR1 | PSAT1 | TG |
| BRAF | FANCE | MAF | PTCH1 | TGFBR1 |
| BRAT1 | FANCF | MAP1A | PTCH2 | TGM1 |
| BRCA2 | FANCG | MAP2K2 | PTH2R | TMEM231 |
| BRIP1 | FANCI | MAPK1 | RAB18 | TMEM94 |
| BSCL2 | FANCM | MASP1 | RAB23 | TNFRSF11A |
| CACNA1A | FBN1 | MDH1 | RAD51 | TNFSF11 |
| CACNA1B | FGFR2 | MEGF8 | RAD54B | TNS1 |
| CACNA2D3 | FGFR3 | MEOX1 | RAD54L | TONSL |
| CC2D2A | FGFR4 | MET | RASA2 | TRIP11 |
| CCDC91 | FGFRL1 | MICALL2 | REPS1 | TRPM4 |
| CD36 | FLNA | MIPEP | REV1 | TSC1 |
| CDC45 | FLNB | MN1 | RNASEH2A | TSC2 |
| CDKAL1 | FLNC | MOCS2 | ROGDI | TTK |
| CDKN1C | FREM1 | MPZL2 | RREB1 | TTN |
| CENPJ | FRMD4A | MTHFR | RYR1 | USH2A |
| CEP135 | FUZ | MTR | SALL1 | USP45 |
| CEP290 | GABBR2 | MUTYH | SALL4 | VANGL1 |
| CEP57 | GATA4 | MYCBP2 | SCN5A | WDPCP |
| CEP63 | GDF5 | MYH3 | SDHB | WDR26 |
| CERKL | GFM1 | MYH7 | SEC24C | WDR35 |
| CERS1 | GH1 | MYH8 | SEC24D | ZIC1 |
| CFAP69 | GLB1 | MYO18B | SENP3 | ZNF341 |
| CHAT | GNA11 | MYO1A | SGCG | ZNF446 |
| CHD2 | GNAS | MYO9A | SHANK1 |  |
| CHKA | GP1BB | MYT1 | SHANK3 |  |
| CHRNE | GPX4 | NANS | SHROOM |  |
| CLIP2 | GTF2IRD1 | NEBL | SHROOM3 |  |
| CLPP | GYS1 | NELFA | SIK3 |  |
| COMT | HAAO | NELL1 | SLC29A1 |  |
| CREBBP | HLCS | NEO1 | SLC30A2 |  |
| CTNND2 | HPSE2 | NF2 | SLC34A2 |  |
| CTSA | IFNG | NID2 | SLC46A1 |  |
| CXXC1 | IFT122 | NIN | SLC7A7 |  |
| DGKD | IL1RN | NOTCH2 | SLMAP |  |
| DMGDH | IMPG1 | NOTCH3 | SLX4 |  |
| CD36 | DNAAF1 | INA | NPHP3 |  |