**Supplementary Table 2: Genetic variants on chromosome 11p15**

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Affected domain(s)**  **T: telomeric**  **C: centromeric** |  | **Affected genes** | **Origin of the pathogenic allele for SRS** | **Reference** |
| T+C | der(4)t(4;11) | *H19*; *IGF2*; *KCNQ1*; *KCNQ1OT1*; *CDKN1C* | Maternal | 1 |
| T+C | dup11p15.5p15.4 | *H19*; *IGF2*; *KCNQ1*; *KCNQ1OT1*; *CDKN1C* | Maternal | 2-4 |
| T+C | der(10)t(10;11) | *H19*; *IGF2*; *KCNQ1*; *KCNQ1OT1*; *CDKN1C* | Maternal | 5 |
| T+C | der(11)t(11;14) | *H19*; *IGF2*; *KCNQ1*; *KCNQ1OT1*; *CDKN1C* | Maternal | 6 |
| T+C | der(11)t(11;15) | *H19*; *IGF2*; *KCNQ1*; *KCNQ1OT1*; *CDKN1C* | Maternal | 7 |
| T+C | der(16)t(11;16) | *H19*; *IGF2*; *KCNQ1*; *KCNQ1OT1*; *CDKN1C* | Maternal | 8 |
| T+C | der(17)t(11;17) | *H19*; *IGF2*; *KCNQ1*; *KCNQ1OT1*; *CDKN1C* | Maternal | 8 |
| Partial T | dup11p15.5 | *H19* | Maternal | 9 (M11221) |
| Partial T | dup11p15.5 | *H19* | Maternal | 10 (S72P) |
| Partial T | del11p15.5 | *H19/IGF2* enhancers; *H19* | Paternal | 11 |
| Partial T | inv(11)(p15.5q21)  including del11p15.5 | *H19/IGF2* enhancers | Paternal | 11 |
| Partial T | Del11p15.5 | *H19/IGF2* enhancers | Paternal | 9 (M6443) |
| C | dup11p15.5p15.4 | *KCNQ1*; *KCNQ1OT1*; *CDKN1C* | Maternal | 12, 13 |
| Partial C | dup11p15.5p15.4 | Partial *KCNQ1*; Partial *KCNQ1OT1*; *CDKN1C* | Maternal | 14 |
| Partial C | del11p15.5 | Partial *KCNQ1OT1* | Paternal | 15 |

**References**

1. Bliek, J. et al. Phenotypic discordance upon paternal or maternal transmission of duplications of the 11p15 imprinted regions. *Eur J Med Genet* **52**, 404-8 (2009).

2. Chiesa, N. et al. The KCNQ1OT1 imprinting control region and non-coding RNA: New properties derived from the study of Beckwith-Wiedemann syndrome and Silver-Russell syndrome cases. *Human Molecular Genetics* **21**, 10-25 (2012).

3. Vals, M. A. et al. Familial 1.3-Mb 11p15.5p15.4 Duplication in Three Generations Causing Silver-Russell and Beckwith-Wiedemann Syndromes. *Mol Syndromol* **6**, 147-51 (2015).

4. Brown, L. A. et al. A cryptic familial rearrangement of 11p15.5, involving both imprinting centers, in a family with a history of short stature. *American Journal of Medical Genetics, Part A* **164**, 1587-1594 (2014).

5. Eggermann, T. et al. Is maternal duplication of 11p15 associated with Silver-Russell syndrome? *J Med Genet* **42**, e26 (2005).

6. Cardarelli, L. et al. Silver-Russell syndrome and Beckwith-Wiedemann syndrome phenotypes associated with 11p duplication in a single family. *Pediatr Dev Pathol* **13**, 326-30 (2010).

7. Eggermann, T. et al. Chromosome 11p15 duplication in Silver-Russell syndrome due to a maternally inherited translocation t(11;15). *Am J Med Genet A* **152a**, 1484-7 (2010).

8. Nakashima, S. et al. Silver-Russell syndrome without body asymmetry in three patients with duplications of maternally derived chromosome 11p15 involving CDKN1C. *J Hum Genet* **60**, 91-5 (2015).

9. Begemann, M. et al. Clinical significance of copy number variations in the 11p15.5 imprinting control regions: new cases and review of the literature. *Journal of Medical Genetics* **49**, 547-553 (2012).

10. Demars, J. et al. New Insights into the Pathogenesis of Beckwith-Wiedemann and Silver-Russell Syndromes: Contribution of Small Copy Number Variations to 11p15 Imprinting Defects. *Human Mutation* **32**, 1171-1182 (2011).

11. Gronskov, K. et al. Deletions and rearrangements of the H19/IGF2 enhancer region in patients with Silver-Russell syndrome and growth retardation. *J Med Genet* **48**, 308-11 (2011).

12. Bonaldi, A. et al. Microduplication of the ICR2 domain at chromosome 11p15 and familial Silver-Russell syndrome. *Am J Med Genet A* **155a**, 2479-83 (2011).

13. Schönherr, N. et al. The centromeric 11p15 imprinting centre is also involved in Silver-Russell syndrome. *J Med Genet* **44**, 59-63 (2007).

14. Xue, Y. et al. Paternal duplication of the 11p15 centromeric imprinting control region is associated with increased expression of CDKN1C in a child with Russell-Silver syndrome. *Am J Med Genet A* (2015).

15. De Crescenzo, A. et al. Paternal deletion of the 11p15.5 centromeric-imprinting control region is associated with alteration of imprinted gene expression and recurrent severe intrauterine growth restriction. *Journal of Medical Genetics* **50**, 99-103 (2013).