



**Supplementary Figure 1.** Flowchart of inclusion criteria.

A total of 330 patients were referred to us for genetic testing for Silver-Russell syndrome (SRS) from 2002 to 2017. Our study included 94 patients without pathogenic CNVs and abnormal methylation levels for nine differentially methylated regions (DMRs) related to known imprinting disorders.

11p15 LOM: loss of methylation on chromosome 11p15; UPD(7)mat: maternal uniparental disomy of chromosome 7; NH-CSS: Netchine-Harblison clinical scoring system; UPD(20)mat: maternal uniparental disomy of chromosome 20; UPD(6)mat: maternal uniparental disomy of chromosome 6; UPD(11)mat: maternal uniparental disomy of chromosome 11.

\*These five pathogenic CNVs were reported in our previous study (Inoue T, Nakamura A, Fuke T, Yamazawa K, Sano S, Matsubara K, Mizuno S, Matsukura Y, Harashima C, Hasegawa T, Nakajima H, Tsumura K, Kizaki Z, Oka A, Ogata T, Fukami M, Kagami M. Genetic heterogeneity of patients with suspected Silver-Russell syndrome: genome-wide copy number analysis in 82 patients without imprinting defects. *Clin Epigenetics* 2017;9:52).