Supplementary material to article by R. Higgins et al. “Uniparental Disomy of Chromosome 2 Unmasks New ITGA6 Recessive Mutation and Results in a Lethal Junctional Epidermolysis Bullosa in a Newborn”

Fig. S4. Mechanism of uniparental disomy occurrence. (A) Chromosomal pattern in parents. (B) The process of meiotic non-disjunction resulting in a trisomy. In most cases this will result in spontaneous abortion or miscarriage. (C) The process of trisomic rescue, where 1 copy of the chromosome is expelled to form a diploid structure. In most cases this results in a normal embryo, but in 30% of cases a uniparental disomy will occur.