



Figure 5. Imprinted clusters associated with the human Beckwith–Wiedemann and pseudohypoparathyroidism imprinting disorders. (A) The expression of imprinted genes at adjacent KCNQ1 and IGF2 imprinting clusters associated with BWS is displayed. Expression patterns from both parental chromosomes in control individuals are indicated. ICRs are indicated in green, which when imprinted, are shown to be DNA methylated (pink hexagon). ICR2 and the antisense KCNQ1OT1 lie within the KCNQ1 locus. The gray arrow connecting ICR1 and ICR2 indicates some kind of regulatory interaction that has been postulated. E, enhancer. (B) The 5' region of the GNAS1 locus is illustrated, a gene implicated in PHP, indicating the parental expression in certain tissues of the different transcripts produced from alternative 5' exons (NESP55, XL, and 1A). The reverse arrow indicates a NESP55 antisense transcript.