

46,XX,inv(2)(p13p23)

**Paracentric** inversion in which breakage and reunion have occurred at bands 2p13 and 2p23. The breakpoint more proximal to the centromere is specified first.

46,XX,inv(3)(q21q26.2)

46,XX,inv(3)(pter→q21::q26.2→q21::q26.2→qter)

**Paracentric** inversion in which breakage and reunion have occurred at bands 3q21 and 3q26.2.

46,XY,inv(3)(p13q21)

46,XY,inv(3)(pter→p13::q21→p13::q21→qter)

**Pericentric** inversion in which breakage and reunion have occurred at bands 3p13 and 3q21. The breakpoint in the short arm is specified first.

### 9.2.11 Isochromosomes

The symbol **i** (not iso) is used for isochromosomes and **idic** for isodicentric chromosomes. The breakpoints in isochromosomes are assigned to the centromeric bands p10 and q10 according to the morphology of the isochromosome. See also Section 9.2.4.

46,XX,i(17)(q10)

46,XX,i(17)(qter→q10::q10→qter)

An isochromosome for the entire long arm of one chromosome 17 and consequently the breakpoint is assigned to 17q10. There is one normal chromosome 17. The shorter designation i(17q) may be used in text but not in the karyotype to describe this isochromosome.

46,X,i(X)(q10)

46,X,i(X)(qter→q10::q10→qter)

One normal X chromosome and an isochromosome for the long arm of one X chromosome.

47,XY,i(X)(q10)

A male showing an isochromosome of the long arm of the X chromosome in addition to a normal X and Y.

46,XX,idic(17)(p11.2)

46,XX,idic(17)(qter→p11.2::p11.2→qter)

An isodicentric chromosome composed of the long arms of chromosome 17 and the short arm materials between the centromeres and the breakpoints in 17p11.2.

46,XX,i(21)(q10)

An isochromosome of the long arm of chromosome 21 has replaced one chromosome 21. There are two copies of the long arm of chromosome 21 in the isochromosome and one normal copy of chromosome 21. Even though there are effectively three copies of the long arm of chromosome 21, the normal chromosome 21 is not designated with a plus sign (+). Note that an alternative description for this same chromosomal rearrangement based on G-banding is found in Section 9.2.17.3 and makes the additional copy of chromosome 21 more obvious.

Complex isochromosomes, including isoderivative chromosomes, are described as derivative chromosomes, see Section 9.2.3.

### 9.2.12 Marker Chromosomes

A *marker chromosome* (**mar**) is a structurally abnormal chromosome that cannot be unambiguously identified or characterized by conventional banding cytogenetics. Numerous terms have been used in the literature to describe markers, including “supernumerary marker chromosomes