Clarification about Chromosomal Formula

The International System for Human Cytogenetic Nomenclature or ISCN has been developed to permit the accurate definition of the chromosomal constitution of an individual.

Conventional cytogenetics

The chromosomal formula is a mean of expressing the result of a karyotype or the chromosomal complement (the whole set of chromosomes in a nucleus):

- In case of Normal karyotype, the number of chromosomes is specified, followed by a comma and the sex chromosomes which exist in the cell.
- In case of chromosomal mutation at the level of autosomes, the number of chromosomes is specified, followed by a comma and the sex chromosome(s) which exist in the cell.
  The latter is followed by a comma and a specification of the chromosome abnormality. Some of the symbols that are used to designate these chromosome abnormalities are:

  - "+" to indicate the presence of a specific additional autosome
  - "–" to indicate the absence of a specific autosome
  - "t" to indicate translocation

- In case of chromosomal mutation at the level of gonosomes, the number of chromosomes is specified, followed by a comma and the sex chromosome(s) which exist in the cell.

  - "O" to indicate a missing sex chromosome
  - Additional X or Y to indicate supernumerary sex chromosomes

Examples:

46,XY	Karyotype of a normal male having 46 chromosomes per cell including one X chromosome and one Y chromosome.
46,XX	Karyotype of a normal female having 46 chromosomes per cell including two X chromosomes.
46, XX, t (1; 18)	Karyotype of a normal female having 46 chromosomes per cell, including two X chromosomes and a translocation between chromosome 1 and chromosome 18.
47,XY,+21	Karyotype of a male with trisomy 21 or Familial Down Syndrome having 47 chromosomes per cell, including one X chromosome and one Y chromosome and an additional copy of Chromosome 2.
45,XO or 45,X	Karyotype of a female with Turner Syndrome having 45 chromosomes per cell, including one X chromosome only; that is, one missing X chromosome.
47, XXY	Karyotype of a male with Klinefelter syndrome having 47 chromosomes per cell, including one X chromosome and one Y chromosome and an additional X chromosome.
45, XX, -13	A female with monosomy 13 or Myeloma Prognosis having 45 chromosomes per cell, including two X chromosomes and a missing copy of Chromosome 13.

References:

- International committee on standardized Genetic Nomenclature for Mice. (2016, June). Rules for Nomenclature of Mouse Chromosome Aberrations. Retrieved from http://www.informatics.jax.org/mgihome/nomen/anomalies.shtml#gband

- Chromosome Disorder Outreach, Inc. (2017, March). Chromosome Disorder out reach. *Introduction to Chromosomes*. Retrieved from http://chromodisorder.org/intro-to-chromosomes