Scripps Center for Learning & Innovation

CHROMOSOMAL ABNORMALITIES

The following are important documentation tips and strategies for this disease/condition as required by the specificity needed in ICD-10:

- Identify the specific numerical and/or structural abnormality such as:

  - *Trisomy 13, 18 &21 - delineate between non-mosaicism, mosaicism, or translocation.*
  - *Partial trisomies &duplications (e.g., whole chromosome non-mosaicism, marker chromosomes in normal individual, polyploidy, etc.).*
  - *Monosomies &deletions (e.g., chromosome replaced with ring, Angelman syndrome, etc.).*
  - *Rearrangements (e.g., chromosome inversion in normal individual, autosomal fragile site, etc.).*
  - *Sex chromosome abnormalities (e.g., Karyotype 45-X, Karyotype 47-XYY, female with more than 3 X chromosomes, etc.).*

- Specify any associated physical condition (e.g., slurred speech, Short stature, spastic gait, etc.) and/or degree of mental retardation (e.g., mild, moderate, severe, etc.).

- List any metabolic disorders (e.g., AAT deficiency, bisalbuminemia, lipodystrophy, etc.).

- Detail any associated deletions or duplications due to unbalanced translocations, inversions, and insertions.

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