

Chromosomal Trisomies

Trisomy is a genetic condition where an individual has three copies of a chromosome instead of the usual two, resulting in a total of 47 chromosomes instead of 46. Common examples include Down syndrome (Trisomy 21), Edwards syndrome (Trisomy 18), and Patau syndrome (Trisomy 13). These conditions can lead to various developmental abnormalities and may affect survival.

ICD-10 CODES

Q90.0 Trisomy 21, nonmosaicism (meiotic nondisjunction)	Q92.0 Whole chromosome trisomy, nonmosaicism (meiotic nondisjunction)
Q90.1 Trisomy 21, mosaicism (mitotic nondisjunction)	Q92.1 Whole chromosome trisomy, mosaicism (mitotic nondisjunction)
Q90.2 Trisomy 21, translocation	Q92.2 Partial trisomy
Q90.9 Down syndrome, unspecified	Q92.5 Duplications with other complex rearrangements
Q91.0 Trisomy 18, nonmosaicism (meiotic nondisjunction)	Q92.61 Marker chromosomes in normal individual
Q91.1 Trisomy 18, mosaicism (mitotic nondisjunction)	Q92.62 Marker chromosomes in abnormal individual
Q91.2 Trisomy 18, translocation	Q92.7 Triploidy and polyploidy
Q91.3 Trisomy 18, unspecified	Q92.8 Other specified trisomies and partial trisomies of autosomes
Q91.4 Trisomy 13, nonmosaicism (meiotic nondisjunction)	Q92.9 Trisomy and partial trisomy of autosomes, unspecified
Q91.5 Trisomy 13, mosaicism (mitotic nondisjunction)	
Q91.6 Trisomy 13, translocation	
Q91.7 Trisomy 13, unspecified	

DOCUMENTATION ACRONYMS

DEEP Diagnosis Elements

Include elements of DEEP in documentation to clinically support chromosomal anomalies.

Diagnosis: Patau syndrome

Evidence: Chromosome translocation, seizures well controlled

Evaluation: Trisomy 13, translocation

Plan: Continue close follow up with pediatric neurology and cardiology

Final Assessment Details

Include DSP for each addressed condition impacting treatment and patient care.

Diagnosis

Chromosomal Trisomy

- Specified Chromosome effected

Status

Complications

- Additional Health factors

Plan

- Treatment of complications
- ADL support if necessary
- Family support if necessary
- Followup with genetics
- Followup with specialists as necessary

BEST PRACTICES & TIPS

- Specificity is key! Always indicate the **specific chromosome anomaly**, any secondary conditions, and use verbiage to solidify the connection between them.
- When documenting a chromosomal defect be sure to **document all health factors** to get a complete picture of the patients' health status.
- DSP should be applied for **chromosome anomalies** as well as for the resulting conditions. Status should be apparent by identifying any required ADL modifications and any treatment or therapies.
- **Avoid using uncertain terms** for confirmed chromosomal defects which include: probable, suspected, likely, questionable, possible, still to be ruled out, compatible with, or consistent with.
- Documentation should **always include DEEP elements** for chromosomal conditions to show clinical evidence as well as any resulting factors and conditions. Incorporate history, tests, imaging, signs and symptoms and document any and all associated treatments.
- Avoid documenting chromosomal anomalies as a "history of" as this **suggests a resolved status** and causes conflict within the documentation.
- **Confirmation** should be found within the documentation representing the complications of a chromosomal condition and any resulting outcomes.



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