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CLINICAL DOCUMENTATION

Monosomies, Translocations and Chromosomal Deletions

A monosomy is a condition where one chromosome in a pair is missing. In a deletion, part of a chromosome is missing. Both monosomies and deletions result in birth defects. A chromosomal translocation is a genetic abnormality where part of one chromosome breaks off and attaches to another, non-homologous chromosome.

ICD-10 CODES

Q93.0 Whole chromosome monosomy, nonmosaicism (meiotic nondisjunction)

Q93.1 Whole chromosome monosomy, mosaicism (mitotic nondisjunction)

Q93.2 Chromosome replaced with ring, dicentric or i sochromosome

Q93.3 Deletion of short arm of chromosome 4

Q93.4 Deletion of short arm of chromosome 5

Q93.51 Angelman syndrome

Q93.52 Phelan-McDermid syndrome

Q93.59 Other deletions of part of a chromosome

Q93.7 Deletions with other complex rearrangements

Q93.81 Velo-cardio-facial syndrome

Q93.82 Williams syndrome

Q93.88 Other microdeletions

Q93.89 Other deletions from the autosomes

Q93.9 Deletion from autosomes, unspecified

Q95.0 Balanced translocation and insertion in normal individual

Q95.1 Chromosome inversion in normal individual

Q95.2 Balanced autosomal rearrangement in abnormal individual

Q95.3 Balanced sex/autosomal rearrangement in abnormal individual

Q95.5 Individual with autosomal fragile site

Q95.8 Other balanced rearrangements and structural markers

Q95.9 Balanced rearrangement and structural marker, unspecified

DOCUMENTATION ACRONYMS

DEEP Diagnosis Elements

Include elements of DEEP in documentation to clinically support a chromosomal disorder.

Diagnosis: Williams Syndrome

Evidence: Confirmed gene deletion of 1.5 to 1.8 Mb on chromosome 7q11.23, hypothyroidism new onset

Evaluation: Williams Syndrome with related hypothyroidism

Plan: Continue followup with cardiology, start synthroid, referral to endocrinology

Final Assessment Details

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

Diagnosis

Chromosomal Anomaly

- 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.

For more resources go to:

HIOSCAR.COM/PROVIDERS/RESOURCES