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CLINICAL DOCUMENTATION

Sex Chromosome Abnormalities

Sex chromosome anomalies are genetic conditions resulting from an abnormal number or abnormal structure of sex chromosomes (X and Y). These anomalies can lead to a range of health conditions, with the most common examples being Turner syndrome, Klinefelter syndrome, XYY syndrome, and Triple X syndrome.

ICD-10 CODES

- **Q96.0** Karyotype 45, X
- **Q96.1** Karyotype 46, X iso (Xq)
- **Q96.2** Karyotype 46, X with abnormal sex chromosome, except iso (Xq)
- **Q96.3** Mosaicism, 45, X/46, XX or XY
- **Q96.4** Mosaicism, 45, X/other cell line(s) with abnormal sex chromosome
- **Q96.8** Other variants of Turner's syndrome
- **Q96.9** Turner's syndrome, unspecified
- **Q97.0** Karyotype 47, XXX
- **Q97.1** Female with more than three X chromosomes
- **Q97.2** Mosaicism, lines with various numbers of X chromosomes
- **Q97.3** Female with 46, XY karyotype
- **Q97.8** Other specified sex chromosome abnormalities, female phenotype
- **Q97.9** Sex chromosome abnormality, female phenotype, unspecified
- **Q98.0** Klinefelter syndrome karyotype 47, XXY
- **Q98.1** Klinefelter syndrome, male with more than two X chromosomes
- **Q98.3** Other male with 46, XX karyotype
- **Q98.4** Klinefelter syndrome, unspecified
- **Q98.5** Karyotype 47, XYY
- **Q98.6** Male with structurally abnormal sex chromosome
- **Q98.7** Male with sex chromosome mosaicism
- **Q98.8** Other specified sex chromosome abnormalities, male phenotype
- **Q98.9** Sex chromosome abnormality, male phenotype, unspecified
- **Q99.0** Chimera 46, XX/46, XY
- **Q99.1** 46, XX true hermaphrodite
- **Q99.2** Fragile X chromosome
- **Q99.8** Other specified chromosome abnormalities
- **Q99.9** Chromosomal abnormality, unspecified

DOCUMENTATION ACRONYMS

DEEP Diagnosis Elements

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

Diagnosis: Turner's Syndrome

Evidence: X chromosome mosaicism confirmed by geneticist, hearing loss new onset

Evaluation: Turner's Syndrome, sensorineural hearing loss

Plan: Urgent referral with ENT, needs hearing test by audiology to evaluate loss

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