ICD-10-CM Documentation
and Coding Best Practices

Guidance on Documentation and Coding for Prader-Willi, Patau's, Edward's and Autosomal Deletion Syndromes

Clinical overview

Prader-Willi syndrome is a rare genetic disorder that results in several physical, mental, and behavioral problems.

Patau's syndrome is a serious rare genetic disorder caused by having an additional copy of chromosome 13 in some or all the body's cells. It's called Trisomy 13.

Edward's syndrome, also called Trisomy 18, is a chromosomal condition associated with abnormalities in many parts of the body.

Autosomal deletion syndromes are defined in two parts:

- Autosomal: involves one of the numbered (i.e., non-sex chromosomes).
- Deletion: an abnormality of DNA that involves missing material. These can range from very small (as little as one base pair) to very large (involving millions of base pairs of DNA).

Prader-Willi syndrome

Q87.11 Prader-Willi syndrome

Causes of this syndrome lie in the genes located in a particular region of chromosome 15. It occurs because certain paternal genes that should be expressed are not, due to:

- Paternal genes on chromosome 15 are missing.
- The child inherited two copies of chromosome 15 from the mother and none from the father.
- There's some error or defect in paternal genes on chromosome 15.

Signs and symptoms can vary among individuals, and may include the following:

For infants:

- Poor muscle tone (hypotonia). Babies may rest with their elbows and knees loosely extended instead of fixed.
- Almond-shaped eyes, narrowing of the head, turned-down mouth, and a thin upper lip.
- Poor sucking reflex due to decreased muscle tone.

Early childhood to adulthood:

- Food craving and weight gain.
- Poor growth and physical development due to an underproduction of growth hormone.
- Mild to moderate intellectual disability, delayed motor development, behavioral problems, and sleep disorders.

Complications of Prader-Willi:

- Obesity-related due to having constant hunger and never feeling full (diabetes, high blood pressure, etc.).
- Inadequate hormone production.
- Other complications such as binge eating and reduced quality of life.

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Patau's syndrome

The cause of Patau's syndrome (Trisomy 13) happens by chance and traces to having three copies of chromosome 13 instead of two.

Signs and symptoms may present as:

- Absence of one or both eyes (anophthalmia).
- Skin missing from the scalp (cutis aplasia).
- Abdominal wall defect, resulting in the intestines being outside the body.

Complications begin almost immediately, and may include:

- Difficulty breathing or lack of.
- Heart failure.
- Seizures.

ICD-10-CM Code information

Trisomy 18 and Trisomy 13

Q91.4	Trisomy 13, nonmosaicism (meiotic nondisjunction)	**Q91.0**	Trisomy 18, nonmosaicism (meiotic nondisjunction)
Q91.5	Trisomy 13, mosaicism (mitotic nondisjunction)	**Q91.1**	Trisomy 18, mosaicism (mitotic nondisjunction)
Q91.6	Trisomy 13, translocation	**Q91.2**	Trisomy 18, translocation
Q91.7	Trisomy 13, unspecified	**Q91.3**	Trisomy 18, unspecified

Edward's syndrome

The cause for most cases of Edward's syndrome (Trisomy 18) results from having three copies of chromosome 18 in each cell instead of the usual two.

Signs and symptoms:

During pregnancy:

- Very little fetal activity.
- A small placenta.

After birth:

- Internal organs forming or functioning differently.
- Issues with cognitive development.
- Weak cry and minimal response to sound.

Complications may include, but not limited to:

- Developmental delay.
- Cardio-respiratory failure.
- Increase risk of neoplasms.

Autosomal deletion syndromes

ICD-10-CM Code information

Q92	Other trisomies and partial trisomies of the autosomes, not elsewhere classified (additional character required)
Q93	Monosomies and deletions from the autosomes, not elsewhere classified (additional character required)
Q95	Balanced rearrangements and structural markers, not elsewhere classified (additional character required)

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Documentation best practices

Provider documentation should include elements such as:

- Clinical features.
- Information of how the individual is developing and functioning.
- Any functional limitations and how they are progressing.
- When the effects of the syndrome affect multiple body systems, evaluate under the appropriate affected system such as musculoskeletal, senses and speech, and neurological.

References

- "ICD-10." Centers for Medicare & Medicaid Services, CMS.gov. http://www.cms.gov/medicare/coding/icd10