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Nursing Management of Patients with Genetic and Congenital Disorders in Adults

Adult Health Nursing

Common genetic conditions presenting in adulthood, inherited disorders, genetic counselling, screening, patient education and nursing responsibilities.

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CHAPTER 21 / ADULT HEALTH NURSING 21. Nursing Management of Patients with Genetic and Congenital Disorders in Adults

1. Basic concepts

A genetic disorder results from variation in genes/chromosomes; congenital means present at birth and may be genetic or environmental. Adult-onset inherited disease can remain unrecognized until adulthood. A three-generation pedigree helps assess family history, age at onset and inheritance.

Pattern	Examples and meaning
Autosomal dominant	Huntington disease, ADPKD, Marfan syndrome; a heterozygous affected parent often has 50% transmission risk each pregnancy.
Autosomal recessive	Sickle cell disease, cystic fibrosis, many thalassaemias; two carriers have 25% affected chance each pregnancy for a typical recessive variant.
X-linked	Haemophilia A/B; transmission depends on sex chromosomes and parental variants.
Multifactorial	Many diabetes, cardiovascular and cancer risks involve multiple genes/environment.

2. Important adult presentations

ADPKD

Definition and pathophysiology: Inherited renal cyst formation, often autosomal dominant.

Clinical features and investigations: Hypertension, flank pain, haematuria and progressive CKD; family history, imaging/genetic testing when indicated.

Management and nursing care: BP management, renal follow-up and counselling about familial risk; assess extrarenal manifestations per specialist advice.

Huntington disease

Definition and pathophysiology: Autosomal dominant CAG repeat disorder causing neurodegeneration.

Clinical features and investigations: Chorea, behavioural/cognitive change, swallowing difficulty; specialist genetic testing.

Management and nursing care: Support mobility, swallowing, mental health, family care and non-directive predictive testing counselling.

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Marfan syndrome

Definition and pathophysiology: Connective-tissue disorder often due to FBN1 variants.

Clinical features and investigations: Tall habitus, skeletal/ocular signs, aortic dilation; clinical criteria, echo and genetic assessment.

Management and nursing care: Aortic surveillance, specialist activity advice and therapy; sudden chest/back pain needs emergency assessment.

Inherited blood and cancer risk

Definition and pathophysiology: Sickle cell/thalassaemia affect haemoglobin; BRCA-related syndromes increase some cancer risks.

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B.SC. NURSING / CHAPTER-WISE STUDY NOTES

Clinical features and investigations: Family history, blood testing or targeted genetic testing; variant interpretation requires expertise.

Management and nursing care: Prevent complications, arrange genetic counselling and risk-based screening. A variant of uncertain significance is not proof of disease.

3. Screening and counselling

Explain purpose, limitations, potential incidental findings, familial implications and consent. Screening estimates risk; diagnostic tests establish specific conditions in context. Protect confidentiality, avoid coercion, facilitate specialist referral and respect decisions about testing. Offer psychosocial support and discuss prevention/surveillance options with genetics team.

4. Important Examination Questions

Long Answer Questions

1. 1. Explain inheritance patterns and nursing role in genetic counselling.

Short Answer Questions

1. 1. Describe ADPKD
2. 2. differentiate genetic and congenital
3. 3. explain pedigree.

Very Short Answer Questions (with answers)

1. 1. What is a pedigree? - Family relationship and trait diagram.
2. 2. Does carrier status always cause illness? - No.

5. Quick Revision

- Genetic risk is not destiny.
- Consent is essential.
- Refer complex results to genetics specialists.

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6. Conclusion

Genetic risk is not destiny. Consent is essential. Refer complex results to genetics specialists.

Practice questions

Choose one best answer. The answer key and explanations follow the questions.

1. What does congenital mean?

A. Present at birth, with genetic or environmental causes possible
B. Always caused by a single inherited gene
C. Always first appearing in adulthood
D. Acquired only after an infection

2. For a typical autosomal recessive variant, what is the affected-child probability in each pregnancy when both parents are carriers?

A. 0%
B. 25%
C. 50%
D. 100%

3. How should a variant of uncertain significance be interpreted?

A. It establishes a definite diagnosis by itself
B. It excludes all inherited risk
C. It is not proof of disease and needs expert interpretation
D. It guarantees future symptoms

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Answer key and explanations

1. A - Present at birth, with genetic or environmental causes possible

Congenital describes presence at birth and does not imply a solely genetic cause.

2. B - 25%

Two carriers have a 25% affected chance in each pregnancy for a typical recessive variant.

3. C - It is not proof of disease and needs expert interpretation

An uncertain variant requires expert interpretation and is not proof of disease.

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