Chapter 18
Human Nervous System Disorders

ABSTRACT

The nervous system (NS) is comprised of nerve cells (neurons), which transfer and process information, and neuroglia (or glial cells), which provide the supportive framework neurons need to function effectively. There are two divisions of the nervous system: central (CNS) and peripheral (PNS). The CNS consists of the brain and spinal cord and forms an intricate network of specialised cells that are responsible for coordinating all bodily functions. The PNS delivers sensory information from peripheral sensory tissues and systems to the CNS and carries motor commands from the CNS to peripheral tissues. This chapter discusses 15 diseases that directly affect the nervous system mostly caused by mutations in a single gene, with others having more complex modes of inheritance. They include Alzheimer’s Disease, epilepsy, essential tremor, familial Mediterranean fever, Friedreich’s ataxia, Huntington’s disease, maple syrup syndrome, Menkes disease, narcolepsy, Parkinson’s Disease, phenylketonuria, Refsum disease, spinal muscular atrophy, tangier disease, and spinocerebellar ataxia.

CHAPTER OUTLINE

18.1 Overview of the Nervous System
18.2 Alzheimer’s Disease
18.3 Epilepsy
18.4 Essential Tumor
18.5 Familial Mediterranean Fever
18.6 Friedreich’s Ataxia
18.7 Huntington’s Disease
18.8 Maple Syrup Urine Disease
18.9 Menkes Disease
18.10 Narcolepsy
18.11 Parkinson’s Disease

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18.12 Phenylketonuria
18.13 Refsum Disease
18.14 Spinal Muscular Atrophy
18.15 Tangier Disease
18.16 Spinocerebellar Ataxia
Chapter Summary

LEARNING OUTCOMES

• Identify each genetic disorder affecting each system
• Outline the symptoms of each disorder
• Explain the genetic basis of each disorder
• Summarize the current therapies available to treat each disorder

18.1 OVERVIEW OF THE NERVOUS SYSTEM

The nervous system (NS) is comprised of nerve cells (neurons) which transfer and process information; and neuroglia (or glial cells) which, twenty times more numerous, provide the supportive framework neurons need to function effectively. There are two divisions of the nervous system: central (CNS) and peripheral (PNS) (Figure 1). The CNS consists of the brain and spinal cord and forms an intricate network of specialised cells that are responsible for coordinating all bodily functions. It is the seat of higher functions like intelligence, memory, learning and emotions and also integrates, processes, and coordinates sensory data and motor commands. The PNS is all neural tissue outside the CNS and delivers sensory information from peripheral sensory tissues and systems like eyes to the CNS. It also carries our motor commands from the CNS to peripheral tissues. Sensory information in the PNS comes from sensory receptors in the eyes, nose, ears, mouth and also from receptors in the skin, muscles, and joints. Motor command information is delivered via the somatic NS which controls voluntary and involuntary skeletal muscle contractions; and the visceral or autonomic NS which controls smooth muscle, cardiac muscles and glandular activity (without conscious control). There several diseases that directly affect the NS with a genetic component—some are due to a mutation in a single gene, while others have more complex modes of inheritance.

18.2 ALZHEIMER’S DISEASE (AD)

According to Alzheimer’s Disease International, it is estimated that there were 46.8 million people worldwide living with dementia in 2015 expected to reach 131.5 million in 2050 (ADI, 2018). The World Health Organization (WHO) states that dementia is a global public health priority as the 7th leading cause of death with approximately 50 million people worldwide with an estimated cost of $818 billion (WHO, 2018). AD accounts for 60–70% of dementia cases worldwide estimated to quadruple by 2050. Currently, approved drugs only temporarily alleviate some of the disease’s symptoms to a limited extent. Global leaders have set a deadline of 2025 for finding an effective way to treat or prevent AD. However, despite