

MEMBRANE, ORGANELLE AND CYTOSKELETAL DISORDERS

OBJECTIVES

After studying this chapter you should be able to:

- list some clinical conditions that arise from specific defects in membranes, organelles and the cytoskeleton;
- relate the symptoms of some of these disorders of membranes, organelles and cytoskeletons to specific defects in their structures and activities;
- discuss the management and treatment of some membrane, organelle and cytoskeletal disorders.

16.1 INTRODUCTION

Membranes are essential for biological activities. A plasma membrane surrounds all eukaryotic cells. Individual organelles are also surrounded by a single and, in some cases, a double membrane or envelope. All these membranes share a common basic structure but differ in their individual compositions that are characteristically adapted to the functions of the cell or organelles in question. Defects in their compositions or structures lead to clinical problems that in many cases are extremely severe or fatal. The recent successes of genome sequencing projects have indicated that as many as 25% of all protein-coding genes may specify the structures of membrane proteins. The shapes and locomotion of cells depend on a highly organized arrangement of fibrous proteins called the cytoskeleton. This is also responsible for the active transport of some materials around the cytoplasm.

This chapter will describe a selected number of diseases or disorders associated with membranes, organelles and the cytoskeleton. These will include diseases associated with defects in nucleocytoplasmic transport; the plasma membrane disorder, cystic fibrosis, and diseases linked to mitochondria, lysosomes, peroxisomes and some cytoskeletal disorders. Many other membrane and organelle associated diseases are not included owing to a lack of space.