Chapter 1 Introduction

1.1 Overview

Genetic and inherited diseases are major medical problems in the world. Many illnesses such as mental disorders, cancers and blood diseases are passed down from one generation to another. Detailed information on the family tree and its analysis are useful to doctors to assist in genetic counselling and in preventing inherited diseases.

Blood diseases such as inherited haemoglobin disorders will undoubtedly cause an increasing health burden in many developing countries (Weatherall, 2005). Improvements in the social-economic status and medical care have resulted in a reduction in ‘treatable’ diseases (infection; malnutrition, etc.). However, there has been an apparent increase in non-treatable diseases such as birth defects. In 1990, more than 20% of infant deaths in Malaysia were attributed to congenital malformations or birth defects, some of which could have been inherited. The exact syndromic diagnosis of babies with multiple birth defects which could not be identified is 22.5% (Thong, 2005).

Thalassaemia is a hereditary disorder that results from insufficient haemoglobin in the red blood cells, which leads to anaemia. According to the Minister of Health, Dr. Chua Soi Lek, any member in a family who is a ‘carrier’ of this disease would be counselled and educated on the risk of conceiving children with thalassaemia (Screening for thalassaemia, 2006).

There is a need for a computerised recording of hereditary diseases and analysis of genetic risk in a family. Doctors are keen to have a computer-aided system to record family information in the form of a family tree showing the relationship of an individual
to his or her family members. The system should allow doctors to document all forms of genetic or inherited diseases in a family for the purpose of medical pedigree analysis.

The system will allow doctors (genetic counsellors) to determine at-risk genetic factors in the family, establish the mode of inheritance, and estimate the genetic risk to other family members and provide summarised information on an individual.

1.2 Statement of Research Problems

In this research, it is important to define the research problem domain and to identify related problems. This will help in determining the focus for the research.

Currently, there are very few pedigree drawing systems that can be used to assist doctors in genetic counselling and in the prevention of inherited diseases. Moreover, the systems that are available do not completely support different types of pedigree analysis and do not use the common or standard human genetic symbols. Thus, many doctors still record hereditary diseases and draw the family tree manually on paper. Manual drawing of the tree is laborious and time-consuming and it becomes difficult when updating a family tree for a big extended family. These weaknesses led to the development of a computer-aided system to facilitate the drawing of pedigree trees.

1.3 Research Objectives

The objectives of this research are:

i. To study the doctor’s requirements analysis of a family tree based on the presence of a genetic condition.
ii. To develop a computer-aided system, PedigreePro, to document information on the family tree to assist doctors in genetic counselling and in the prevention of inherited diseases.

1.4 Scope of Research

Interviews were conducted with the medical professionals to determine the scope of the information required for genetic counselling and in preventing inherited diseases. The interviews helped to determine how genetic counselling should be conducted with the aid of the family tree.

Further discussions were held with a clinical geneticist from the Department of Paediatrics, Faculty of Medicine, University of Malaya. A follow-up discussion with a genetic counsellor provided further understanding of the domain requirements.

The method to be used to transform the family tree information into a computer-aided system was determined after the domain study. This was followed by a study of other pedigree drawing systems available overseas.

1.5 Project Schedule

The research commenced in December 2004 and was completed in May 2006. The development of PedigreePro, starting from the requirements gathering phase until the implementation and evaluation phases took 16 months. The development schedule is shown in Figure 1.1.
1.6 Summary of Dissertation

This dissertation consists of six chapters. The contents of each chapter are briefly described below.

Chapter 1 discusses the background of the research and identifies the research problem. The problem statements help to determine the objectives of this research. This is followed by the definition of the scope of the research and a plan of the project schedule.

Chapter 2 reviews the literature on the recording of hereditary diseases and analysis of genetic risks in a family. This is followed by a study and review of the pedigree analysis elements. In addition, other similar systems available in the market are reviewed.

Chapter 3 focuses on the interview with a domain expert and follow-up discussion with a genetic counsellor to further understand pedigree analysis and domain analysis. It also describes the methodologies used in the research and development of the system.

Chapter 4 describes in detail the system requirements analysis and design. This chapter also describes the system architecture, database design, user interface design, and genetic symbols design for the development of PedigreePro.
Chapter 5 explains the development tools used and the coding aspect of the system. It also explains the tests conducted on PedigreePro to ensure that it is free of errors and bugs.

Chapter 6 discusses the problems encountered during the research and the strengths and limitations of PedigreePro. It also includes suggestions for future enhancements and ends with a conclusion.