

SINDROMES: A PROGRAM FOR COMPUTER-ASSISTED DIAGNOSIS OF MALFORMATION SYNDROMES

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ABSTRACT

We have developed a computer program, called SINDROMES which was designed to assist in the differential diagnosis of the more common dysmorphic syndromes. The program is menu-driven and simple to use, not demanding any computer experience. It is written in COBOL and contains two main data bases: a "Syndromes Data Base" with 316 syndromes and their clinical features and a "Characteristics Data Base" containing 671 clinical features hierarchically organized and with code numbers that permit their rapid identification. The main feature of the program is the capability of generating differential diagnostic lists of syndromes based on the clinical characteristics observed in a patient. Program routines also allow the listing of characteristics of a given syndrome, the listing of any data base, and easy expansion or modification of the data bases. The program can be run in microcomputers under the CP/M operational system (64 Kbytes minimum) or MS/DOS operational system (256 Kbytes minimum) and only needs two disk drives for 5 1/4" or 3 1/2" double-face diskettes.

INTRODUCTION

Approximately 0.7% of all newborns suffer from multiple malformations ascertainable at birth (Smith, 1982). It is essential that a precise syndrome diagnosis be made to guide treatment, to establish clinical prognosis and to direct genetic

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counselling. During the past twenty years great efforts have been made in the delineation of discrete syndromes with homogeneous etiology and pathogenesis. These efforts have already allowed the description of more than 1500 known syndromes, and this number has increased constantly (Gouvernet *et al.*, 1985). Since no physician can remember so many diseases, several of which are very rare, syndrome diagnosis has become very time consuming, demanding hours of literature search (Feingold, 1982; Winter and Baraitser, 1984; Buyse, 1984). This situation is ideal for the application of computer assistance and indeed several software packages have been developed for this purpose (Aymé and Gouvernet, 1982; Buyse, 1984; Bachman *et al.*, 1984; Winter *et al.*, 1984; Schorderet and Aebischer, 1984, 1985; Gouvernet *et al.*, 1985; Center for Birth Defects Information Services, 1985; Computer Power Pty, 1986; Schorderet, 1987). All these programs are expensive and demand relatively sophisticated hardware such as hard disks and, in one case, a laser disk system.

We have developed a computer system for aid in syndrome diagnosis that, although much more limited in scope than most the ones referred to above, presents some distinct advantages:

- 1) The program demands simple hardware. It can be run in any microcomputer with more than 64 Kbytes RAM in the CP/M or 256 K RAM in the MS/DOS operational systems, needing only two drives for 3 1/2" or 5 1/4" floppy diskettes. A printer is highly recommended.

- 2) The program has a limited scope and concentrates on the syndromes listed in two standard texts: "*Recognizable Patterns of Human Malformations*" (Smith, 1982) and "*Clinical Atlas of Human Chromosomes*" (De Grouchy and Tourleau, 1984). Thus it is easy to work from the differential diagnosis lists given by the computer by using these books, which have all the pertinent pictures. Because of its simplicity, this program should also prove very useful in the teaching of dysmorphology.

- 3) In contrast to other software available, the data base also includes chromosomal disorders. Thus it can be used by clinicians with limited resources, in order to permit a more rational use of the cytogenetics laboratory.

- 4) The program will be distributed free of charge to all interested agencies.

TECHNICAL DESCRIPTION

The program was written in COBOL (*Common Business Oriented Language*; Microsoft, 1982; 1984). It is menu driven and very easy to use and does not demand previous experience. It contains a managing system and two main data bases: a **Characteristics Data Base** and a **Syndromes Data Base**.

The **Characteristics Data Base** contains 671 symptoms and signs hierarchically organized in 17 main groups. Each of the 671 clinical characteristics has a six digit code. The first two digits identify the main subgroup while the others indicate the anomaly with increasing specificity. For instance, osseous syndactyly of the fingers has the code 152641: the two first digits identify the subgroup limb anomalies, the digit 2 specifies upper limbs, the 6 specifies fingers, the 4 specifies syndactyly and the 1 osseous syndactyly.

The **Syndromes Data Base** contains all the syndromes, sequences and ruptures listed in the two source texts, listed in alphabetical order or, in the case of chromosomal disorders, by the numerical order of the chromosome involved. Each syndrome received a code number.

There is also a **Syndrome characteristics Data Base** which contains the clinical description of each syndrome.

OPERATIONAL FEATURES

As mentioned above the program is menu-driven and very easy to use without any training. After booting, the menu gives a series of choices. The most used and indeed the *raison d'être* of the program is the "**Diagnostic Consultation**". After choosing this option, the user is prompted to enter the characteristics of the patient. Up to ten characteristics can be used in a search. However, experience has shown that searching based on more than three characters is not very productive. For instance in a patient with mental retardation, microcephaly, microphthalmia, cleft lip and palate, congenital heart disease and post-axial polydactyly it would not be worthwhile do enter all these features. It would be more fruitful to choose two or three of these characteristics (e.g. cleft lip and polydactyly) and produce a reasonable differential diagnosis list. If the list were too large, other characteristics could be added to reduce it's size. Since the algorithm involves sequential searches, it is time-saving to first indicate the rarer features. The program then outputs a differential diagnosis list, with a printing option. It is important to emphasize that the syndromes are listed in numerical order and not in order of likelihood. The user then has the option of looking up the characteristics of the listed syndromes in the appropriate reference texts or to retrieve them from the database using the "**Syndrome Characteristics**" option in the menu.

DISCUSSION

The ability of the clinician to interpret all the signs and symptoms of the patient and reach a diagnosis cannot be replaced by any machine. Thus, in our

opinion, the syndromologist does not need computer programs that can make diagnoses *for* him, but rather programs that help him as a memory aid. This is the philosophy of **SINDROMES**, which is a simple program designed to give a list of syndromes (differential diagnosis) based on the clinical characteristics of a patient. From the differential diagnosis list the dysmorphologist will then pick the one that most closely matches his patient based on personal experience or utilizing the source texts of Smith (1982) and De Grouchy and Tourleau (1984). For those that do not have these texts, the program has a routine that permits listing of all the clinical features of a given syndrome. It cannot be overemphasized that the clinical skill of the consulting dysmorphologist is essential for good operation of the program, not only in making the final diagnosis but also in selecting the characteristics which will be entered for the patient. As mentioned previously, the program is simple to use and needs only unsophisticated hardware. The fact that only syndromes from two standard texts are used, simplifies the analysis of the differential diagnosis list. On the other hand, **SINDROMES** has the drawback of containing only a relatively small number of syndromes. This was a necessity imposed by the need to keep all information on two floppy disks. If this capacity were exceeded we would have to use a hard disk, which would be expensive, or to swap diskettes frequently, which would be inconvenient. Anyway, the program offers as an option a simple routine to alter the data bases by modifying existing data or adding new characteristics or new syndromes. This should permit anyone to expand the program to his own needs or customize it to a specific subgroup of syndromes of individual interest, such as skeletal dysplasias, ophthalmic syndromes etc. We are currently using this routine to update the program according to the 1988 edition of *Smith's Recognizable Patterns of Human Malformation* (Jones, 1988).

Any geneticist interested in a copy of **SINDROMES** should send us two 5 1/4" double sided diskettes together with a description of their operational system and a check for 10 BTN's (or US\$15.00) to cover photocopying and mailing.

RESUMO

Desenvolvemos um banco de dados computadorizado, denominado "SINDROMES", destinado a auxiliar o diagnóstico diferencial das síndromes disfórmicas mais frequentes. Trata-se de um sistema simples que não exige do usuário conhecimentos em informática. Escrito em linguagem COBOL, contém um arquivo de Síndromes com as principais características clínicas, ordenadas hierarquicamente e com números de código que permitem sua rápida identificação. A principal rotina de programa gera listas de Síndromes para o diagnóstico diferencial, a partir de características clínicas selecionadas dos pacientes. É também possível listar as características clínicas de síndromes escolhidas para comparação com os pacientes. Rotinas de programa também permitem alterar cada um dos arquivos possibilitando a

correção, atualização e ampliação dos mesmos com a inclusão de novas síndromes. O sistema pode ser utilizado em microcomputadores sob sistema operacional CP/M (Mínimo 64 Kbytes) ou MS/DOS (Mínimo de 256 Kbytes) e duas unidades de disquetes face dupla de 5 1/4" ou 3 1/2".

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