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Diagnostics of malformation syndromes using an available computer program*

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Increased attention to the malformation syndromatology is motivated by the importance of accurate diagnosis and its necessity in genetic counselling. Given the great number of malformation syndromes, it is logical to make use of computers in their ability perform differential diagnostics. The aim of our present examinations was to assess the benefit that an experienced pediatrician can obtain from an available program. The "Syndrome Program" of R. Winter and M. Baraitser was applied. Using coded symptoms, the computer produces a short list of possible diagnoses, which can then be assessed by the user. References and syndrome features can be recalled and compared with those in undiagnosed cases.

The diagnoses of 100 different syndromes were controlled. The method applied yielded very good results. The computer diagnosis of malformation syndromes is a helpful way of supplying information. It may promote the standardization of diagnostic practice, international cooperation and epidemiologic studies in different regions.

In recent years, paediatric syndromatology has gained importance in the field of clinical practice. Dealing with the problem is important due to the following aspects:

1. Exact diagnosis has always been the prime task of clinical work.

2. In syndromatology, the delineation of the independent character of syndromes has been the first step in recognizing the nature of pathological processes for various diseases.

3. In further phases of progress, practical results have evolved in this field. Nowadays, in connection with most syndromes, more and more precise genetic councelling can be given. Progress has been made in the treatment of numerous syndromes, moreover, even prevention in some cases has been made possible [2, 4].

Clearly, in this field of clinical knowledge, we are confronted with the task of looking for logical connections among numerous data. Thus, it is obvious that we should make use of computers.

When we tried to use the help obtained by computers in the examination of clinical pictures of malformations [3, 5, 6, 7, 8, 9] we wanted to investigate the diagnostical problems to be solved from purely clinical aspects. It seemed worthwhile to examine — in addition to diagnostic help — what aid could be offered by this

* Dedicated to Professor Walter Teller on the occasion of his 60th birthday.

rather complicated field for a general practitioner paediatrist in information.

Methods

Equipment through the help of the Genetic Expert Committee of the International Paediatric Association, we have obtained the computer program, elaborated by Winter & Baraitser [9], from the clinical work team of the Hospital of Great Ormond Street of London.

The program was obtained — at our request — on a floppy diskette sent by post and could be used on a microcomputer having a hard disk data recorder with a high output. Our equipment, used for this purpose, was a Commodore PC20.

The attached instructions were enough to use it practically without any special mathematical-technical expertness after a few days practice. Naturally the more our experience increased, the quicker the rapidity and effectivity of the decisions improved.

Program of the study: consisted of studying the usefulness of computer aided diagnostics, as well as of practicing it. We, further, performed the following additional work using our computer program:

A. By using the syndromatology found in textbooks, special monographs and case reports, we wanted to see whether the required syndrome was included or not in the list of the possible diagnoses.

B. By using the data of our already diagnosed cases, occurring in our case records and in everyday practice, we attempted to ascertain whether by computer diagnostics the same, already known, diagnosis could be obtained or not.

C. We have used it in diagnosing our own cases, in which a specific diagnosis has not been decided upon. In the majority of these cases, our diagnosis was only a tentative general definition, and figured usually as multiplex, general disorder in our reports.

D. Based upon description sent by other health centres, we have made counselling service.

Results

ad. A. In the first group the diagnoses of 26 syndromes were made, by the help of a computer, based upon case records described in books or other reference sources.

The diagnoses of the cases examined in this group, when a computer was used with good results, are shown on Table I.

Table I

Computer successfully used in the diagnostic of syndromes published in textbooks, monographs, case studies

Apert	Holt—Oram
BBB (Opitz)	LADD
Beckwith—Wiedemann	Langer—Giedion
Bloom	McKusick metaphys. dyspl.
Chegiak—Higashi	Nail-patella
Cleft lip-ectrodact.	Oculo-Dento-digital
Coffin—Lowry	Oro-facial-digital
Contractural arachnodact.	Refsum
Crouson	Rutledge
Deafness with ichthyosis	Sturge—Weber
EEC	Tricho—Rhino—Phalangeal
Fabry	Weaver
GM_2 gangliosidosis	Zellweger

TABLE II

Computer successfully used in the diagnostic of syndromes previously diagnosed in our Department

Alkaptonuria	Gaucher	Prader—Willi
Alport	Glycogenosis type III.	Prune belly
Arthrogryposis	Hunter	Recklinghausen
Cleidocranialis	Hurler	Roberts
Cockayne	Incont pigmenti	Rubinstein—Taybi
Conradi	Ivemark	Sanfilippo
De Lange	Jeune	Schwachman
EEC	Klippel—Feil	Sclerosis tub.
Ehlers—Danlos	Lowe	Seckel
Ellis-van Creveld	McCune—Albright	Silver—Russel
Fanconi pancytopenia	Melkerson—Rosenthal	Sotos
Farber	Milroy	Thanatophor dyspl.
Fetal alkohol	Morquio	Usher
Fibrodyspl. ossif. progr.	Osteogen. imperf.	VATER
Fraser	Poland	Waardenburg

ad. B. In the control of previously diagnosed diseases, in 45 cases, the same diagnosis was obtained or an even more precise or specific one could be made by using the list of symptoms or the literary data given by the computer. The diagnoses of these cases are seen on Table II.

In groups A and B there were only 6 cases in which the otherwise certain diagnosis was not confirmed by the computer. The syndromes in which the computer failed to give a correct diagnosis are seen on Table III.

TABLE III

Syndromes in which the diagnostic use of the computer was unsuccessful

Olier
Friedreich atax
Blackfan—Diamond
Wiscott—Aldrich
Wilson
Lesh—Nyhan

ad. C. Computer diagnostics was especially helpful in the diagnoses of cases not known by us. In this group, the diagnoses of several syndromes could be seen, despite the fact that they were not previously diagnosed. This was the first case in which we were able to recognize these syndromes.

The syndromes, first verified with the help of a computer, are seen on Table IV.

ad. D. We performed examinations for other centers as well. We obtained the clinical data and photographs of the cases and were informed of their supposed diagnosis only after having enclosed our own evaluation. This was the way when we did not obtain the cooperation of the Paediatric Institute of Moscow. This methodology was considered helpful, in the cases that were evaluated in this manner; both by those in need of counsel-

TABLE IV

First time identified syndromes by the assistance of the computer aided diagnostic in our department

Alopecia with antibody defic.	Schwartz—Jampel
Corpus callosum agenesia	Smith—Lemli—Opitz
Dubowitz	Tollner
Ichthyosis-cataracta	Werner
Kaufman—McKusick	X-linked arthrogryposis
Leber amaurosis	X-linked cataract

Failure in the use of computer in unknown syndromes
No of cases: 7

ling, and by our group. We are of the opinion that, by computer aided diagnostics, the diagnostic activity of other centres can also be helped.

DISCUSSION

Summarizing our observations, we are able to say that in our work, there is no reason to believe that professional or clinical experience will ever be replaced by computer aided diagnosis, it is only a clinical stand-by. It is similar to a good textbook which opens at the very page which is needed. I consider the system designed by Winter and Baraitser [9] especially good, as this program, while looking for a diagnosis, follows the same steps that the clinician would; while creating a diagnosis in the traditional way. In addition to helping medical practice, increasing professional knowledge, promoting expertness in syndromatology, there are great perspectives in the field of international cooperation by using identical and similar programs. The fact that, without proper genetic specialization, we were able to use a computer aided diagnostic system may give stimulation for recommending this process as suitable for widerange, practical, clinical application.

There are, however, opponents to computer aided diagnosis. One of the most important counter-arguments is that there is some danger that the method used by unauthorized hands can lead to abuse. Alas, the only reply may be that, similarly to other events in medicine; for instance in the use of antibiotics and other drugs, some sort of abuse cannot be avoided. However, in the course of time, they may be eliminated, and faulty practice cannot discredit the method itself.

REFERENCES

1. Bankier A, Haan EA, Danks DM: Syndrome Identification Research within the Royal Children's Hospital Parkville, Victoria 3052, Australia, Hospital Edition, p. 1985. 11.
2. Bergsma D: Malformation Syndromes Birth Defects Series Excerpta Medica, Amsterdam, 1975
3. Gal E Gal I: Human Congenital Malfor-

mations. The Design of a Computer-aided Study. Butterwords London, 1975
4. Harner PS: Practical Genetic Counselling. Ed. Wright, Bristol, 1981
5. XVIII. International Congress of Pediatrics, Honolulu-Hawaii. Abstracts of Scientific Presentations. No: 156, 163, 354, 716, 1047; 1986.
6. Leiber B: Dofonos ein "Deutsches Syndrom-Identifikations und Informations System". Medizin 22: 864, 1974
7. Leiber B, Olbrich G: Die Klinische Syndrome. Urban und Schwarzenberg, München, Vols I., II. 1981.
8. Schoderet D, Aebischer P: Microcomputer Based Differential Diagnosis of Malformation Patterns. Arch Dis Childh 60: 248, 1985
9. Winter RM, Baraitser M, Douglas JM: A computerized data base for the diagnosis of rare dysmorphic syndromes. J Med Genet 21: 121, 1984

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