SYNDROC ON THE WEB

USER'S MANUAL

by

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1. Introduction

Diagnosing human malformation syndromes is difficult for two reasons : the rate of appearance of new syndromes and the number of potential diagnoses. SYNDROC has been written to partially overcome these difficulties by building a database describing all the frequent dysmorphic syndromes. The first version of SYNDROC appeared in 1983 (Schorderet D & Aebischer P : Microcomputers and malformation syndromes. Arch Dis Childh 59:1004,1984)At that time, SYNDROC was written in TurboPascal. Since then, it has been updated at different occasions, depending on the time left by my other interests.

Internet and the WEB system have been a wonderful revolution for various reasons: easy access, fast upgrades, worldwide distribution, reduced costs, and maybe the most important of all, ability to establish unlimited links to other databases. For these reasons, I've been rewriting SYNDROC to use this new mode of computing. I would like to take this opportunity to thank the University Hospital of Lausanne (the CHUV) for housing my database and allowing you to access this site for free.

2. Accessing SYNDROC on the WEB

If you are reading this file, it probably means that you have obtained a username and password to access SYNCDROC. Access to SYNDROC will be granted to all specialists in medical genetics, so there is no need to give you password to other colleagues. There is also a simple reason not to give this password to your friends: the CHUV provides free access to their WEB site, but I have to show them that this program is used by the medical genetics community. If not, they may not see the reason to keep this site.

3. Starting

Open your WEB browser and type Syndroc's URL (http://syndroc.hospvd.ch :8004/). By the way, I'm using Explorer as navigator; other programs may not work the same.

User ID: type your username

Password: type your password

4. SYNDROC on the WEB

4.1 Choosing a search option :

Choose « Syndrome from signs » if you want to search for syndromes matching specific signs

Choose « Signs of a syndrome » if you want to see a description of a syndrome

Then click OK

4.1.1. Syndrome consultation

4.1.1.1 Entering signs

The screen is split in three windows, one on the left side (the list of findings), one in the upper right (the introduction window) and one in the lower right part of the screen (the choosing window).

Position your cursor in the introduction window and type « a ». Because the sign you entered is not unique, a list of all the signs starting with « a » is given in the choosing window. Choose the desired sign by clicking on it. If your introduction is unique and corresponds to a sign in the database, it is directly transferred to list of findings. Because the description of dysmorphic syndromes is difficult, authors have used numerous words to describe their findings. Sometimes these words do indeed describe different features, sometimes they are synonymous. I've tried where it was possible to include these synonyms as part of SYNDROC's database. Sometimes, traits used to describe specific syndromes have not been entered. So it may be possible that you will try to introduce findings not contained in SYNDROC's database. In that case, you will see a message telling that this sign doesn't exist. This may be due to the fact that this sign is indeed not part of SYNDROC's thesaurus. It may also be that you have made a mistake in typing this trait. In that case, it is a good approach to only type the first few letters and let SYNDROC proposes a list of signs. For example, introducing « prenatal on » will make SYNDROC proposes « prenatal onset growth retardation » which is what you want. This feature is not new if you've been using SYNDROC already.

You can type as much as 10 signs. You can delete signs, etc. You can search with 3 signs for example and then add two more signs for another search. When doing so, you need to position the cursor at the next available box in the list of findings, or else the 4th sign you enter will overwrite the first.

SYNDROC on the WEB will search the selected database with the list displayed in the list of findings' box.

4.1.1.2. Results of a search

4.1.1.2.1. Descriptive algorithm

SYNDROC on the WEB provides a list of all the candidate syndromes ordered by the number of introduced shared signs. The first column indicates the name of the syndrome, the second its OMIM number and the third the number of signs from your introduction list shared by this syndrome.

Clicking on each underlined or blue name can trigger a new action. For example, clicking on the name of the diagnosis calls a new window describing the features of the diagnosis. Use your browser's BACK option to return to the previous screen. Clicking of the OMIM number will call OMIM web site and position you on that syndrome. From there, you have access to Medline, sequence data, mutation databases, etc. Use the BACK option to come back to SYNDROC on the WEB.

When you are in the description of a diagnosis, clicking on any sign will list a differential diagnosis of the trait.

Sometimes, diagnoses don't have OMIM number (for example foetal rubella). In that case, I've tried to link to a reference database.

4.1.1.2.2. Pseudo-Bayesian algorithm

When you are done searching SYNDROC's database with the descriptive algorithm, go back to the introduction window and choose the pseudo-Bayesian approach by clicking the box « Bayesian ». Except for the fact that the proposed diagnoses are not listed with the number of signs in common but rather with a CC, the options are identical.

4.1.1.3 Printing reports

There is no special printing capacity in SYNDROC on the WEB. You have to use your browser's printing option.

4.1.2 Chromosome consultation

SYNDROC on the WEB allows you to search two databases. The program functions are identical in both databases.

4.2 Choosing the database

SYNDROC on the WEB has two databases, a syndrome and a chromosome database. By default, the syndrome database is selected.

4.3 Choosing an algorithm

4.3.1 The descriptive algorithm

SYNDROC on the WEB has two search algorithms. In the descriptive algorithm, each syndrome is checked for the presence of the signs you typed. The syndromes are then listed accordingly to the number of positive signs. If several syndromes have the same number of signs in common, they are displayed by alphabetical order.

4.3.2 The pseudo-Bayesian algorithm

Using the same signs, you can search both databases with the pseudo-Bayesian algorithm by clicking on the « Bayesian » box. In this algorithm, each sign has a specific value (weight) for each diagnosis. The diagnoses are ranked by the sum of all values obtained from each sign and listed with their Coefficient of Certitude (CC). You can imagine, although it's completely wrong statistically, that the CC provides a sort of probability of this syndrome being the correct diagnosis. It's statistically wrong because the frequency of the different signs in each diagnosis, the frequency of these signs in the general population and the frequency of the syndromes are all unknown, or known with such a poor certitude that these numbers cannot be used statistically.

Anyway, even if the numbers are not statistically correct, they can be used to obtain a good estimate.

After having entered several signs, each diagnosis of the database has a CC which can go from 0 to 1. SYNDROC on the WEB displays only a fraction of these diagnoses. The number of proposed diagnoses is a function of the highest CC : all the diagnoses having a CC comprises between 100% and 50% of the highest CC will be displayed.

When searching the chromosome database with the pseudo-Bayesian algorithm, one has to remember that the description of most of these chromosomal aberrations is based on few reports and therefore, the different members of the Bayesian equation may be even more wrong than with the syndrome cases.

5. Help from you

As you may imagine, keeping such databases up-to-date takes a lot of time. If you have corrections, additions, comments, etc. do not hesitate to contact me.