SYNDROC

Medical Diagnosis System

USER MANUAL

Internet version — syndroc.com

Version 2.2

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About Dr Schorderet

1. About this manual

SYNDROC is a clinical diagnostic aid for rare diseases and dysmorphic syndromes. You describe a patient by selecting clinical findings, and SYNDROC returns diagnoses that best fit the selected findings. The platform also includes an interactive teaching module.

Important SYNDROC is a clinical memory aid and teaching tool. It does not make a diagnosis. The displayed percentages are internal ranking scores, not probabilities that a patient has a particular disease. Results require clinical interpretation and, when appropriate, molecular, cytogenetic or other confirmatory testing.

1.1 Who this manual is for

This manual is intended for clinicians, geneticists, pediatricians, trainees and other authorized users of the internet version of SYNDROC. It does not describe system administration or server maintenance.

1.2 Language

The internet interface and this manual use English. If you encounter a French label or message on syndroc.com, please report it because it is not intended to remain in the user interface.

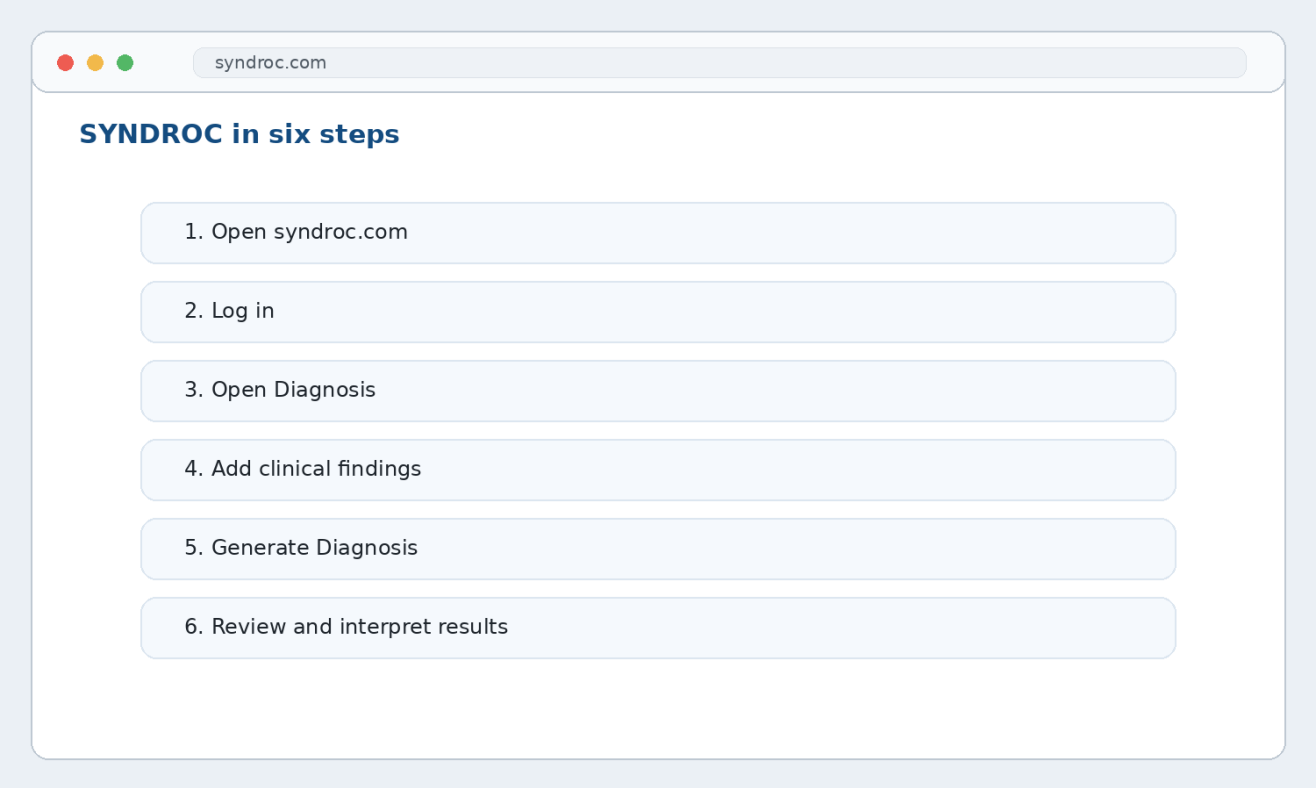


Figure 1 — The ordinary user workflow on syndroc.com.

2. Accessing SYNDROC on the internet

2.1 Open the website

1. Open a modern web browser such as Safari, Chrome, Edge or Firefox.
2. Go to https://syndroc.com.
3. The SYNDROC sign-in page should appear.

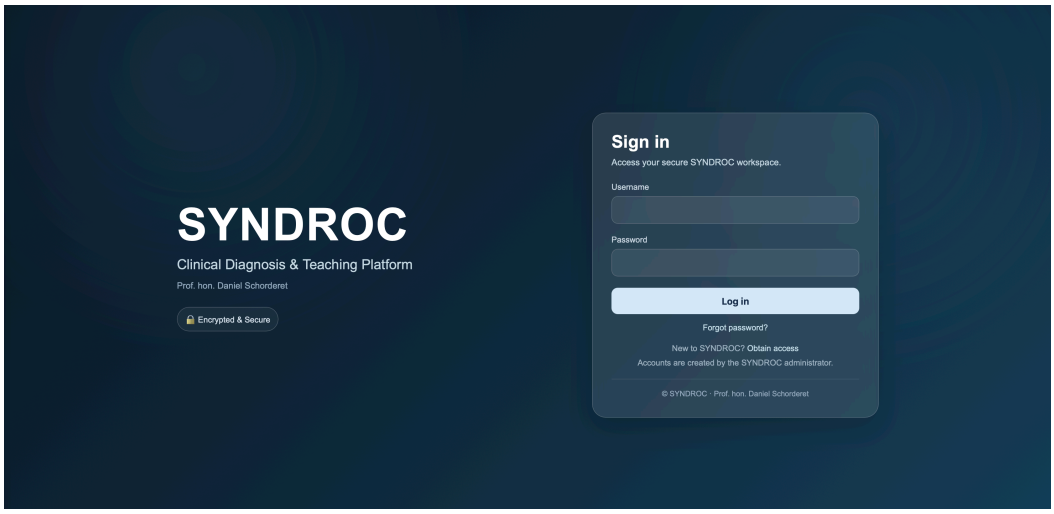


Figure 2 — SYNDROC sign-in screen. Interface wording reflects the current internet version.

2.2 Sign in

1. Enter your Username.
2. Enter your Password.
3. Click Log in.

After successful authentication, SYNDROC opens your secure workspace.

2.3 End your session

Use Logout when you finish, especially on a shared or institutional computer. Closing the browser alone should not be used as a substitute for signing out.

3. Accounts, access requests and passwords

3.1 Requesting access

If you do not yet have a SYNDROC account, click Obtain access on the sign-in page. Complete the form with your first name, last name, university or institutional affiliation, degree and email address. The request is reviewed by the SYNDROC administrator.

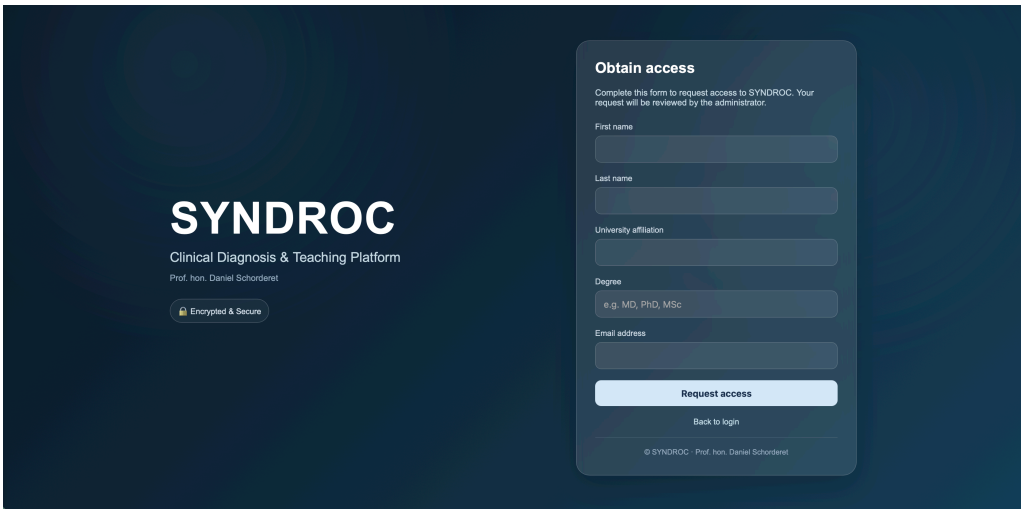


Figure 3 — Obtain access form for a new SYNDROC user.

3.2 Creating an account

Depending on the access route provided to you, you may be invited to choose a username and password and to verify your email address with a 6-digit code. An invitation code may activate an account immediately; otherwise administrator approval can be required.

3.3 Password rules

- Use at least 12 characters.
- Choose a password that is different from any temporary or default password.
- Do not share your password with colleagues.
- Do not reuse your institutional password if your institution advises against password reuse.

3.4 Forgotten password

1. Click Forgot password?
2. Enter your username or email address.
3. Request a reset code.
4. Enter the 6-digit verification code sent by email.

5. Choose and confirm a new password of at least 12 characters.

3.5 Account status messages

Message	Meaning
Invalid credentials	The username or password is incorrect.
Account awaiting administrator approval.	The account is not active yet.
Account rejected or disabled.	The account cannot currently sign in.
New password required	A temporary/default password must be replaced before continuing.

4. Guided demonstration

After you sign in, SYNDROC can guide you through its diagnosis and teaching features. The demonstration uses sample findings and a teaching case so you can see the workflow before entering your own findings.

4.1 Start or restart the demonstration

On your first visit, choose Start guided demo on the welcome screen. To watch it again later, use the Demo button on the SYNDROC home page. Spoken explanations accompany the highlighted controls and pointer.

4.2 Follow the diagnosis example

The tour opens Differential Diagnosis, shows the database and algorithm choices, and enters five example findings: supravalvular aortic stenosis, mental deficiency, midfacial hypoplasia, bifid scrotum and ectrodactyly. It generates a ranked result using the Pseudo-Bayesian algorithm, opens a diagnosis, and then compares the Descriptive ranking. It also shows the Common Findings and Specific Findings controls and demonstrates how to search for a diagnosis by name.

4.3 See the teaching example

The tour then opens the Interactive Training Platform, shows its difficulty levels and presents a Level 1 example case. The example illustrates how clinical findings are grouped by frequency. It does not require you to submit an answer or change your training score.

4.4 Control the tour

When Click to continue appears, click once to advance. In the diagnosis part of the tour, pressing any key stops the demonstration. You can restart it from the Demo button on the home page. The teaching part displays its own prompts for continuing.

5. The SYNDROC home page

After signing in, the home page is the starting point for the functions available to your account.



Figure 4 — SYNDROC home page for an ordinary user.

Area	Purpose
Diagnosis	Enter clinical findings and generate a differential diagnosis.
Interactive Training Platform	Practice dysmorphic diagnosis and self-training.
About	Information about SYNDROC and its author.
Logout	End the current session.

The page also displays the software version and the date of the most recent medical database update.

6. The diagnosis application

The diagnosis application is the core of SYNDROC. It can generate a differential diagnosis from selected clinical findings or help you inspect a diagnosis that you already know by name, synonym or OMIM number.

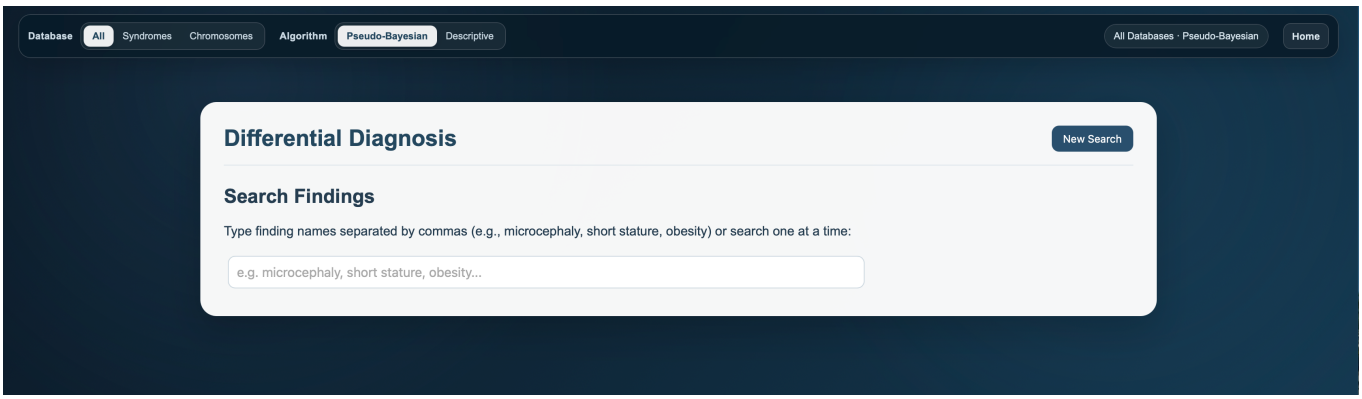


Figure 5 — Diagnosis screen showing database choice, algorithm and differential diagnosis window.

6.1 Select the database

Option	Searches
Both Databases / All Databases	Syndromes and chromosomal anomalies together.
Syndromes Only	Named syndromes only.
Chromosomes Only	Chromosomal anomalies only.

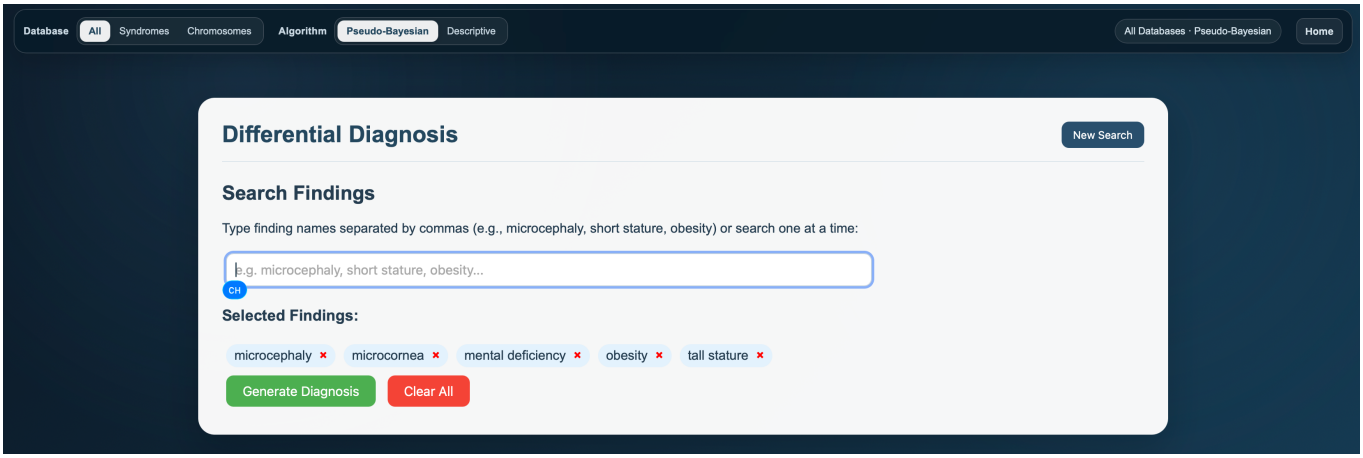
6.2 Select an algorithm

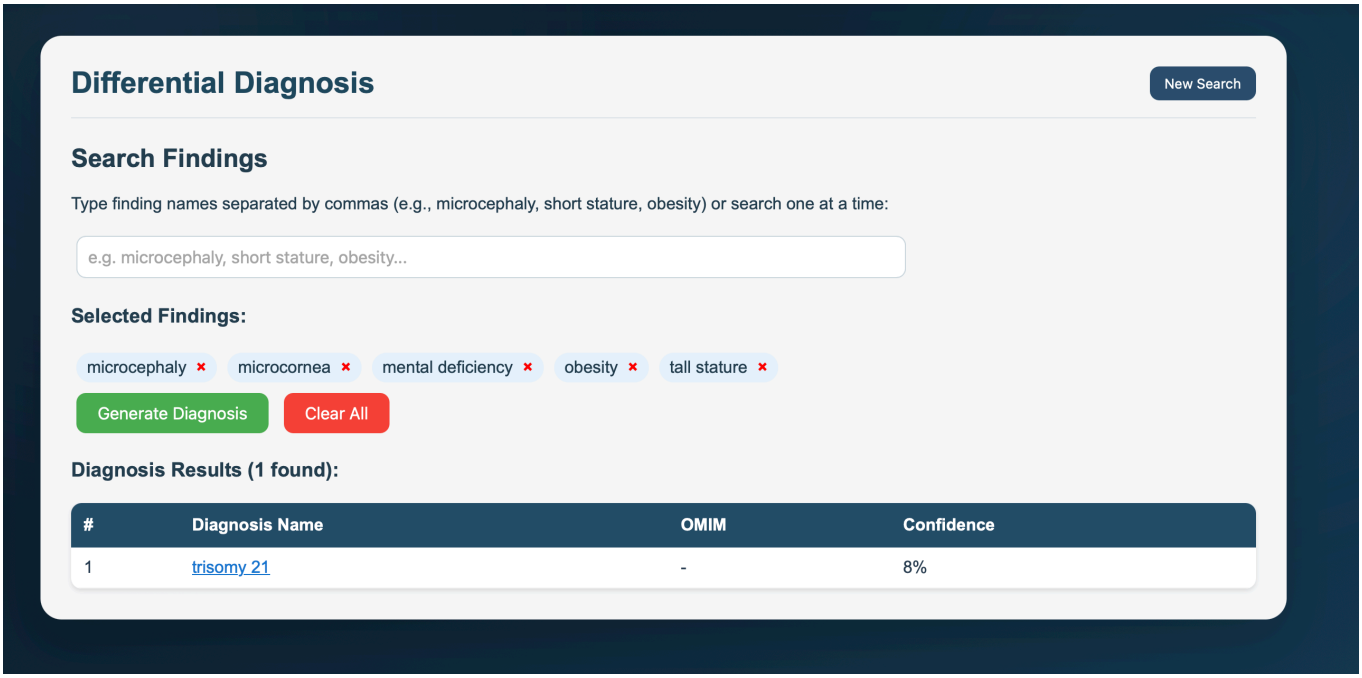
Pseudo-Bayesian is the normal starting point. Descriptive is useful when you want to broaden the differential or emphasize direct overlap between the selected findings and the diagnoses.

6.3 Enter clinical findings

1. Start typing a finding.
2. Choose the appropriate suggestion from the list.
3. Repeat for each relevant finding.
4. Remove a finding if it was selected by mistake.

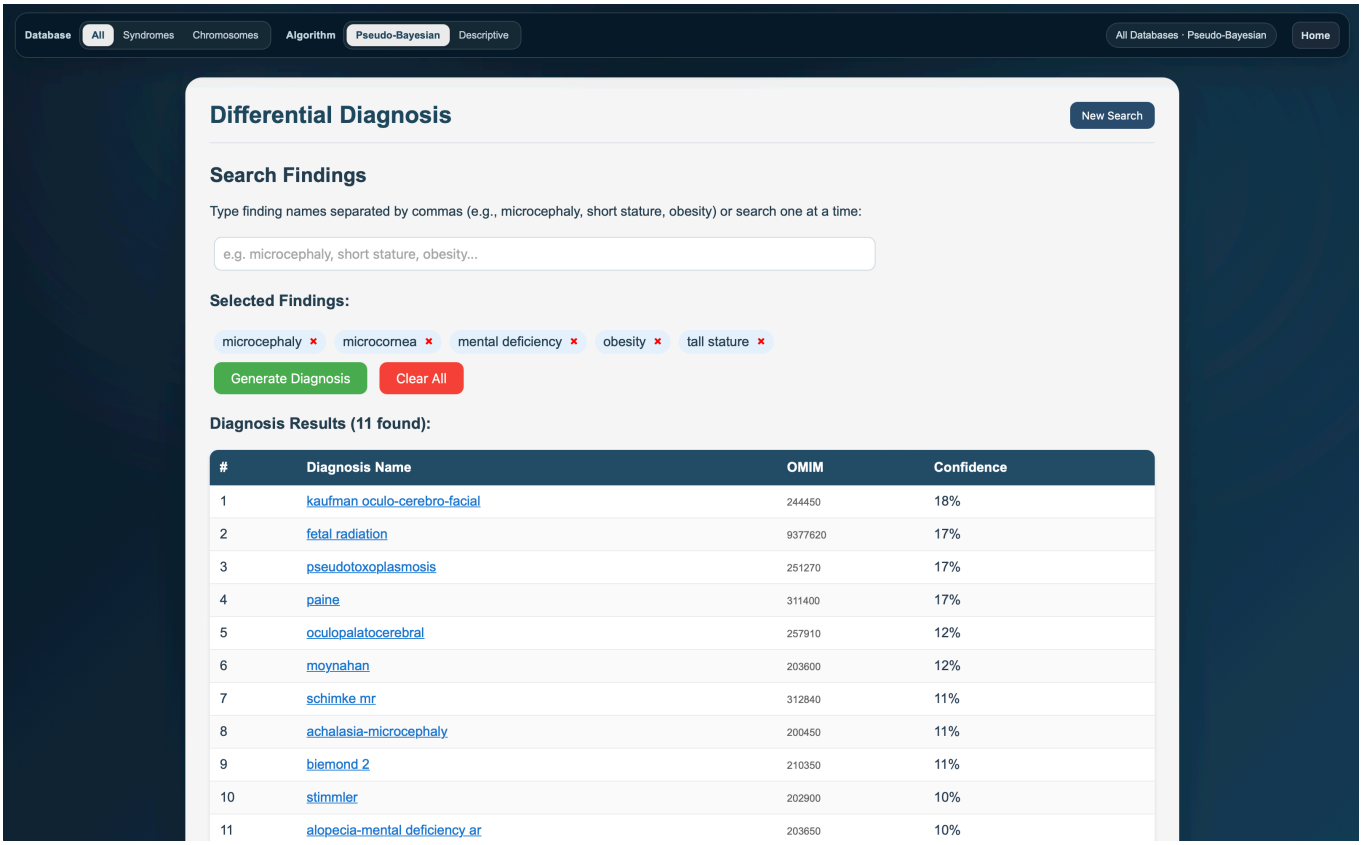
Use specific, well-observed findings whenever possible. Adding many uncertain or redundant findings can make the ranking less useful.





6.4 Generate a differential diagnosis

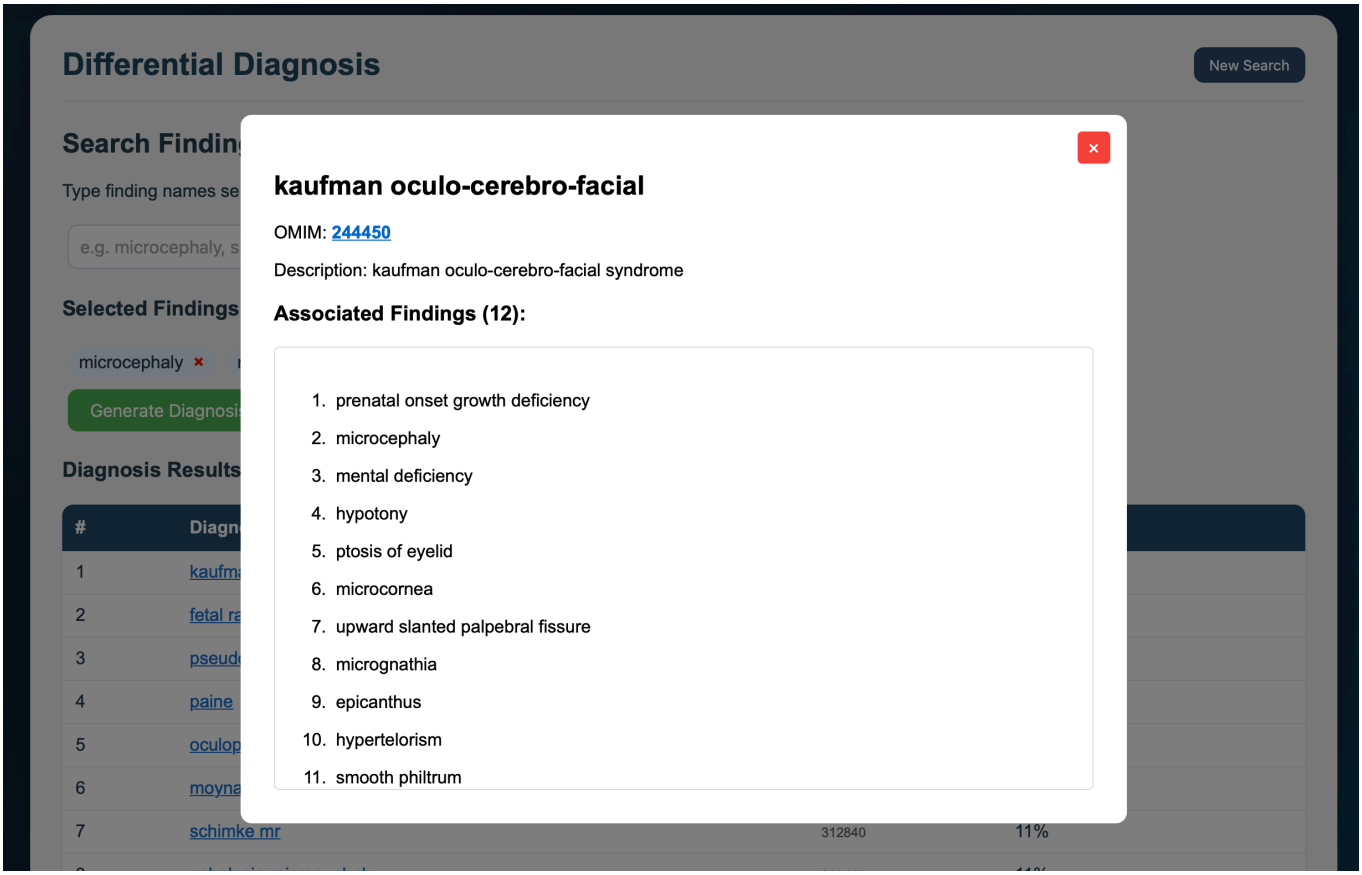
Click Generate Diagnosis. SYNDROC displays a ranked result list. Review the highest-ranked diagnoses but also consider lower-ranked diagnoses when the phenotype is incomplete, age-dependent or atypical.



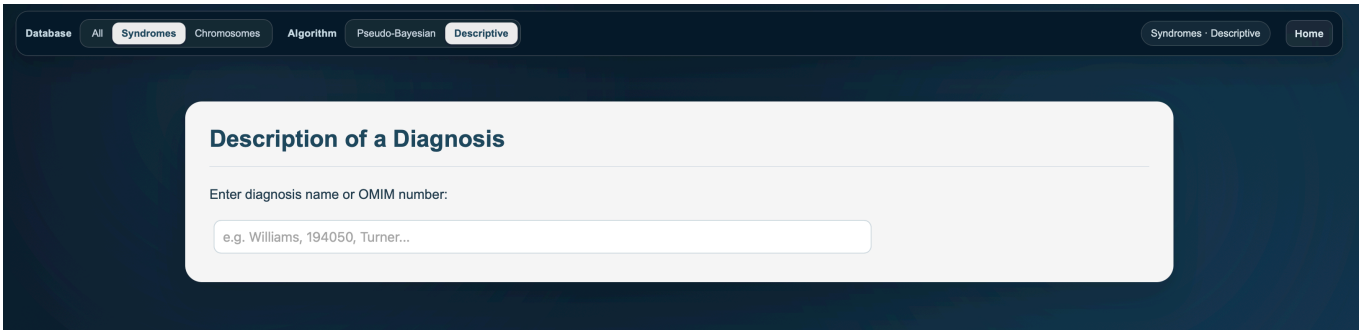
Do not interpret the score as risk A score such as 84% is an internal ranking value within the current search. It does not mean that the patient has an 84% probability of the disease.

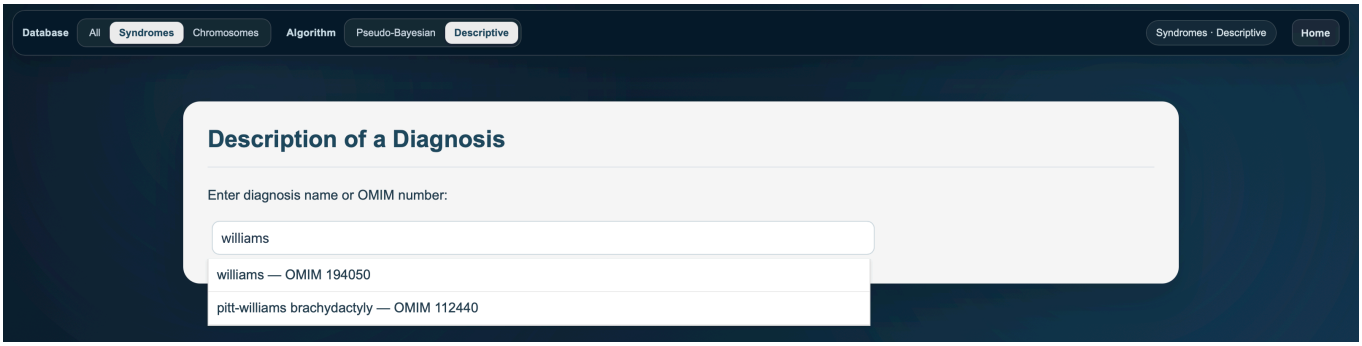
6.5 Inspect a diagnosis

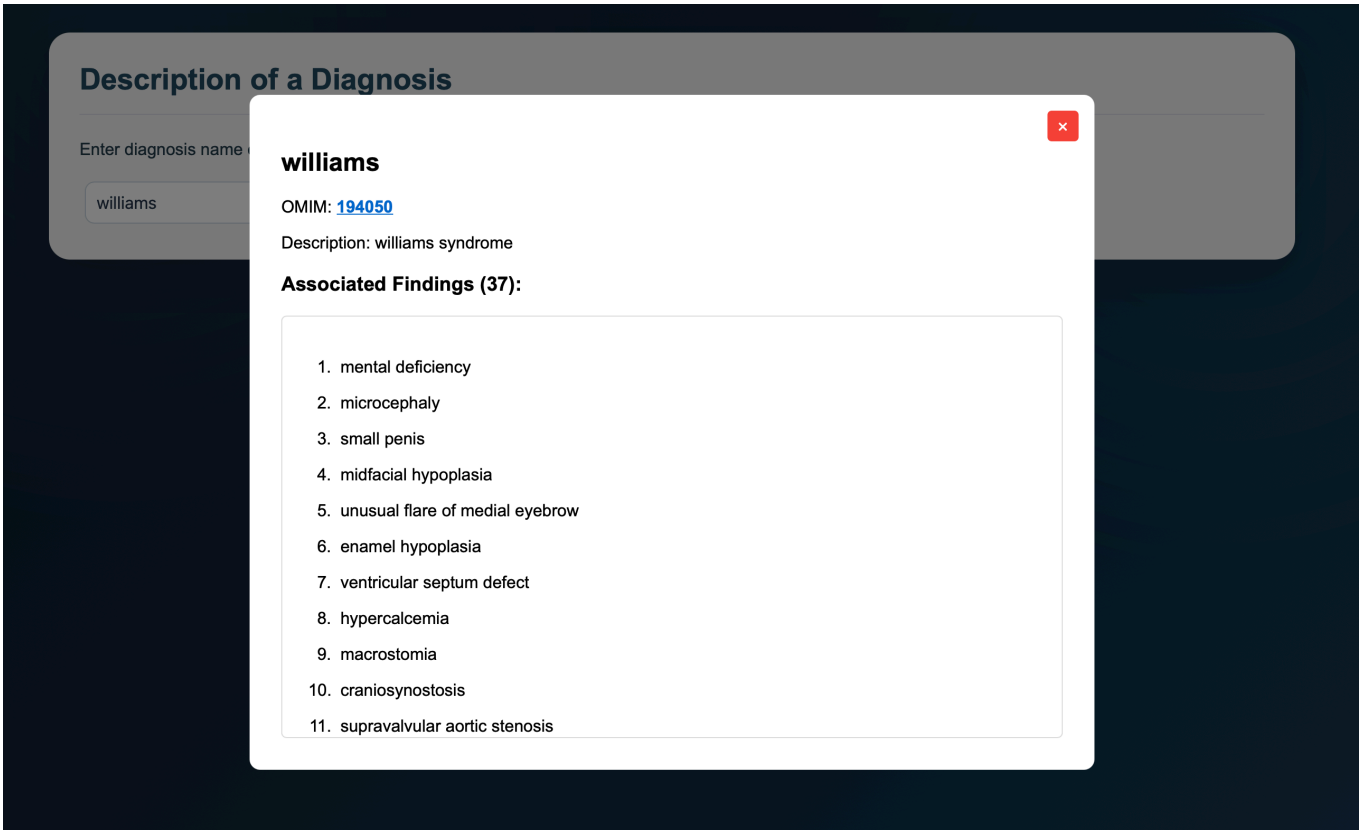
Open a diagnosis to review its description, OMIM information when available, and the clinical findings associated with it. Compare the listed findings with the patient phenotype and verify the diagnosis independently when needed.



7. Description of a diagnosis




Database
All
Syndromes
Chromosomes
Algorithm
Pseudo-Bayesian
Descriptive
Syndromes · Descriptive
Home
Description of a Diagnosis
Enter diagnosis name or OMIM number:
williams
williams — OMIM 194050
pitt-williams brachydactyly — OMIM 112440


Description of a Diagnosis
Enter diagnosis name
williams
williams
OMIM: 194050
Description: williams syndrome
Associated Findings (37):
1. mental deficiency
2. microcephaly
3. small penis
4. midfacial hypoplasia
5. unusual flare of medial eyebrow
6. enamel hypoplasia
7. ventricular septum defect
8. hypercalcemia
9. macrostomia
10. craniosynostosis
11. supravalvular aortic stenosis

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7. Understanding the two algorithms

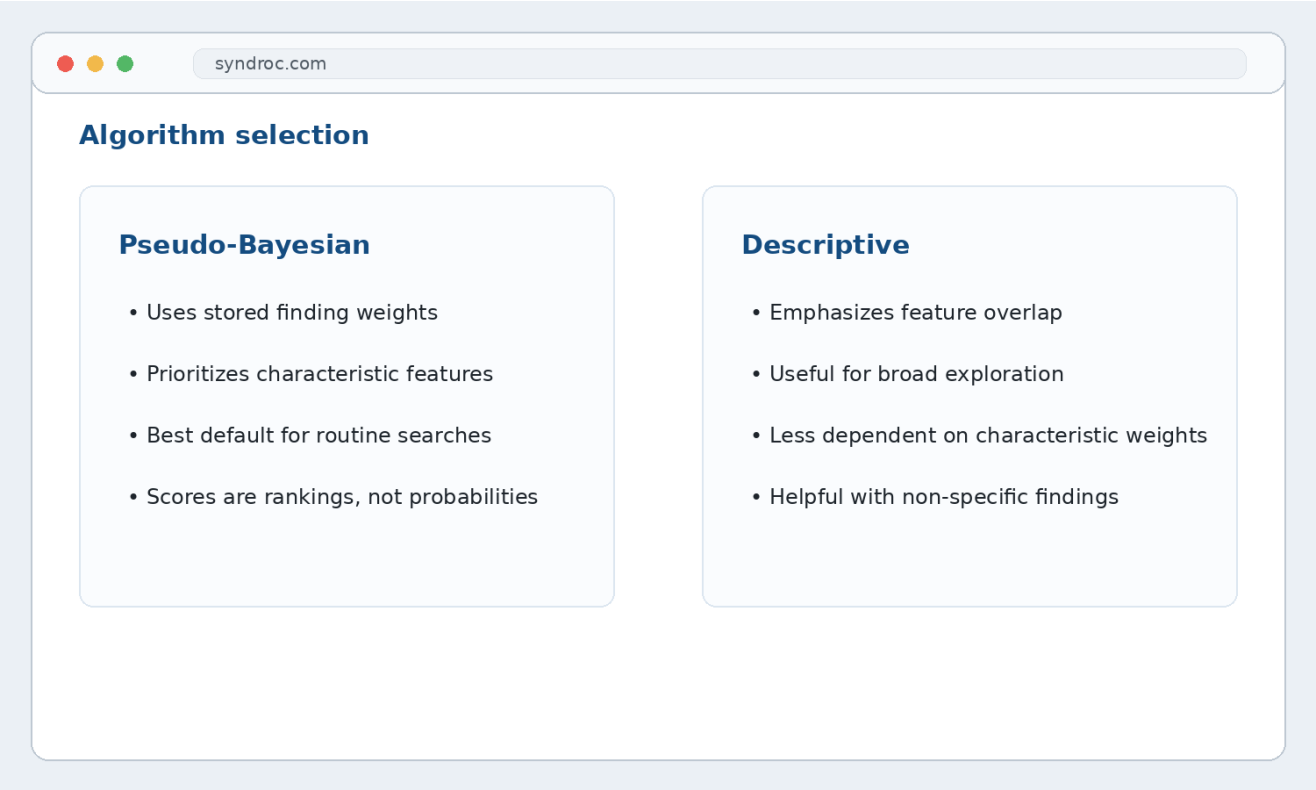


Figure 6 — Pseudo-Bayesian and Descriptive modes serve complementary purposes.

7.1 Pseudo-Bayesian mode

This mode uses the stored weights of diagnosis–finding associations. Highly characteristic findings therefore contribute more strongly than non-specific findings. The result is a clinically oriented ranking, not a formal posterior probability.

- Recommended as the default mode for routine use.
- Useful when the selected findings include distinctive features.
- Can rank a diagnosis lower when only a small proportion of its expected phenotype is represented.
- Does not convert the displayed score into a real disease probability.

7.2 Descriptive mode

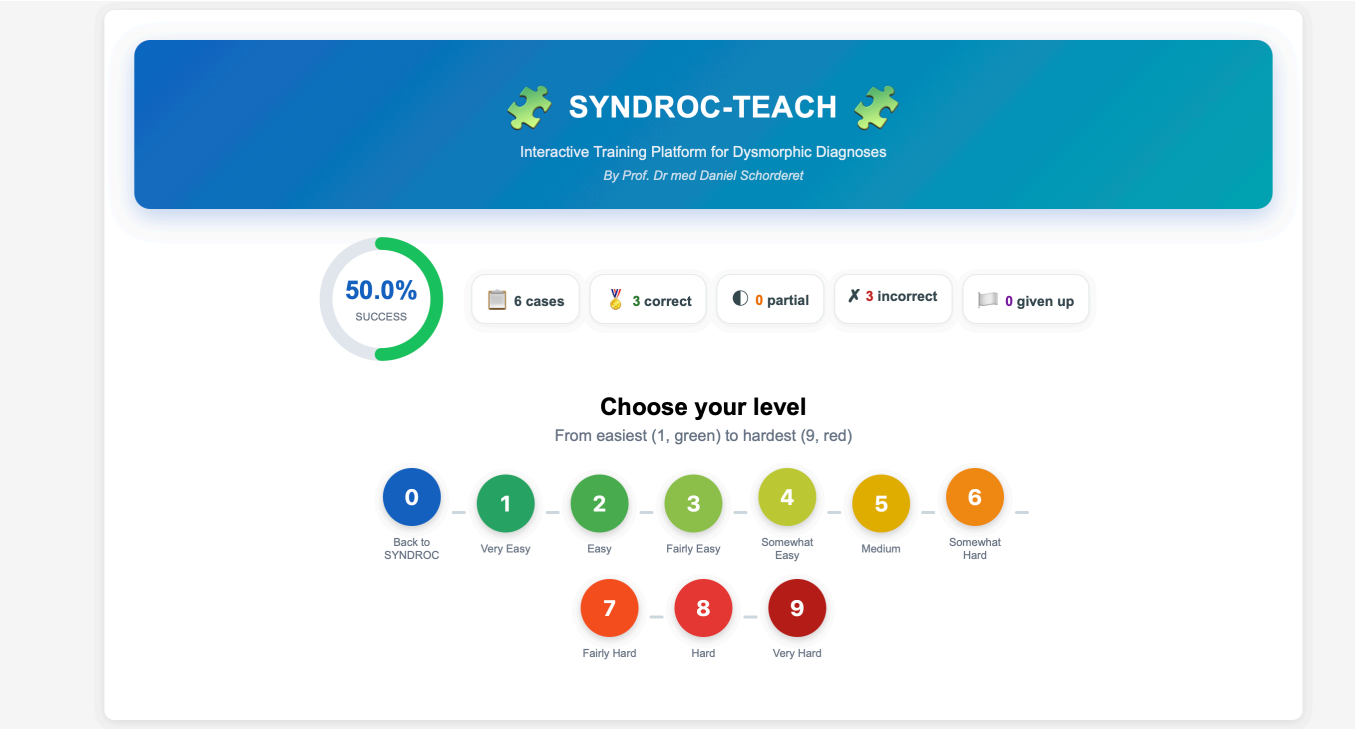
This mode emphasizes overlap between your selected findings and the findings stored for each diagnosis. It is useful when the phenotype is broad, when findings are non-specific, or when you want a wider differential diagnosis. Diagnoses with the same number of matching selected findings can receive identical descriptive scores; these scores are not disease probabilities.

7.3 Practical strategy

A useful workflow is to begin with Pseudo-Bayesian mode, review the result list, and then repeat the same findings in Descriptive mode if the first result is unexpectedly narrow or does not fit the clinical impression.

8. The teaching module

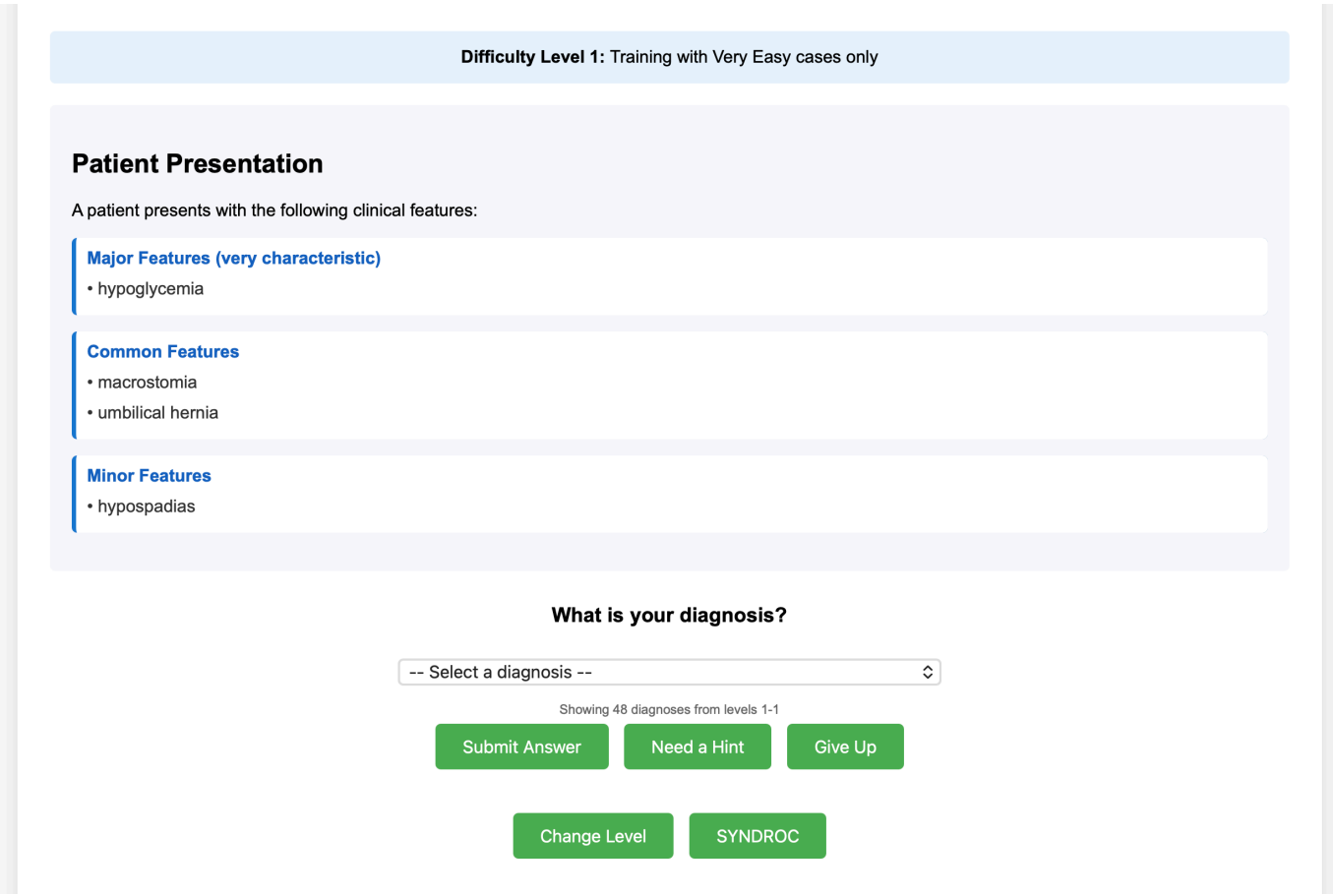
The Interactive Training Platform for Dysmorphic Diagnoses is available from the SYNDROC home page. It is designed for self-training and teaching based on the same diagnostic knowledge base used by the main application.



Choose level, 1 is the easiest, 9 the hardest.

Teach will arbitrarily select a syndrome from which it will select several findings.

You should choose between the proposed diagnoses.



Once you have selected an answer hit the button Submit Answer.

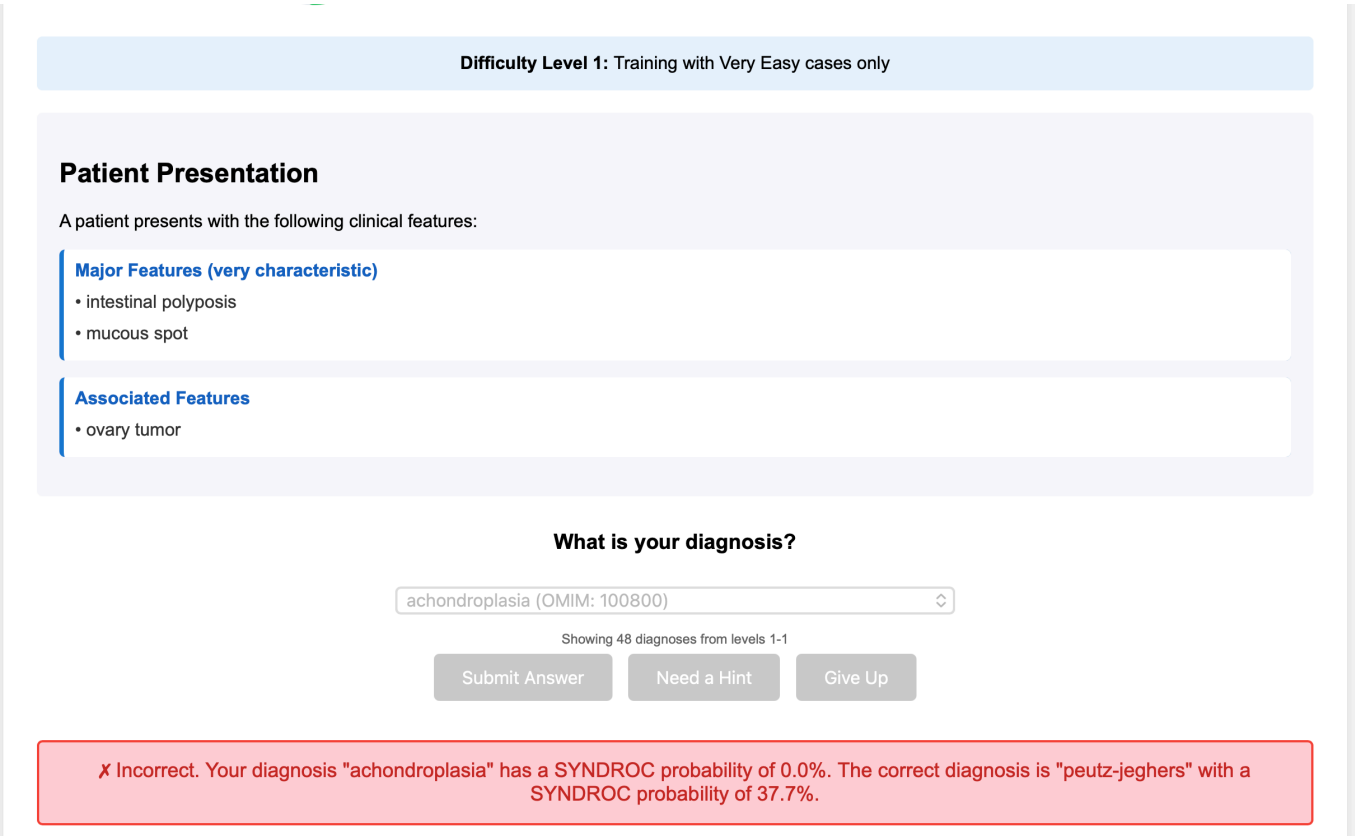


Figure 7 — Representative teaching screen with a case, selected findings and answer field.

If your answer differs from the expected diagnosis, SYNDROC compares it with the findings shown in the case. Because the findings are sampled, another diagnosis may also fit well. Review the feedback before moving to the next case.

8.1 Working through a case

1. Review the findings presented in the training case.
2. Formulate your differential diagnosis before entering an answer.
3. Submit your diagnosis.
4. Review the feedback and the expected diagnosis.
5. Continue with additional cases to consolidate pattern recognition.

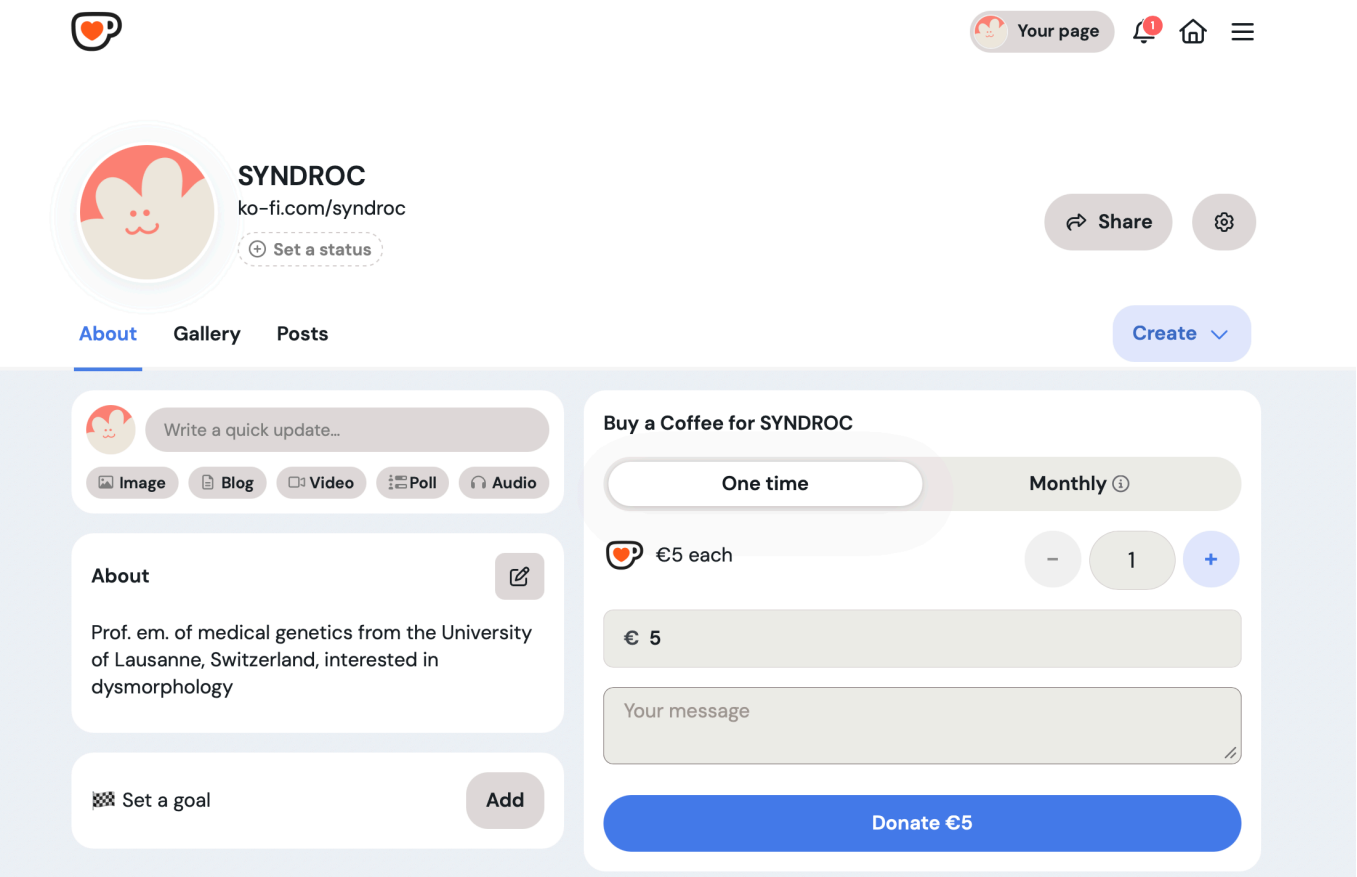
8.2 Difficulty and scores

Cases can be selected according to difficulty. Training scores and progress can be associated with your user account so that progress can be followed across sessions.

Teaching data The teaching module is intended for training. Do not enter patient-identifying information into teaching fields.

9. Helping SYNDROC

SYNDROC is available as a free clinical and teaching resource. If you find it useful and wish to support its maintenance and development, use the Support SYNDROC option on the home page. Contributions are voluntary and do not affect access to the clinical functions.



10. Data, privacy and secure use

SYNDROC is a diagnostic knowledge system, not an electronic patient record. The safest way to use the platform is to enter phenotype information only and to keep patient identity in the clinical record system used by your institution.

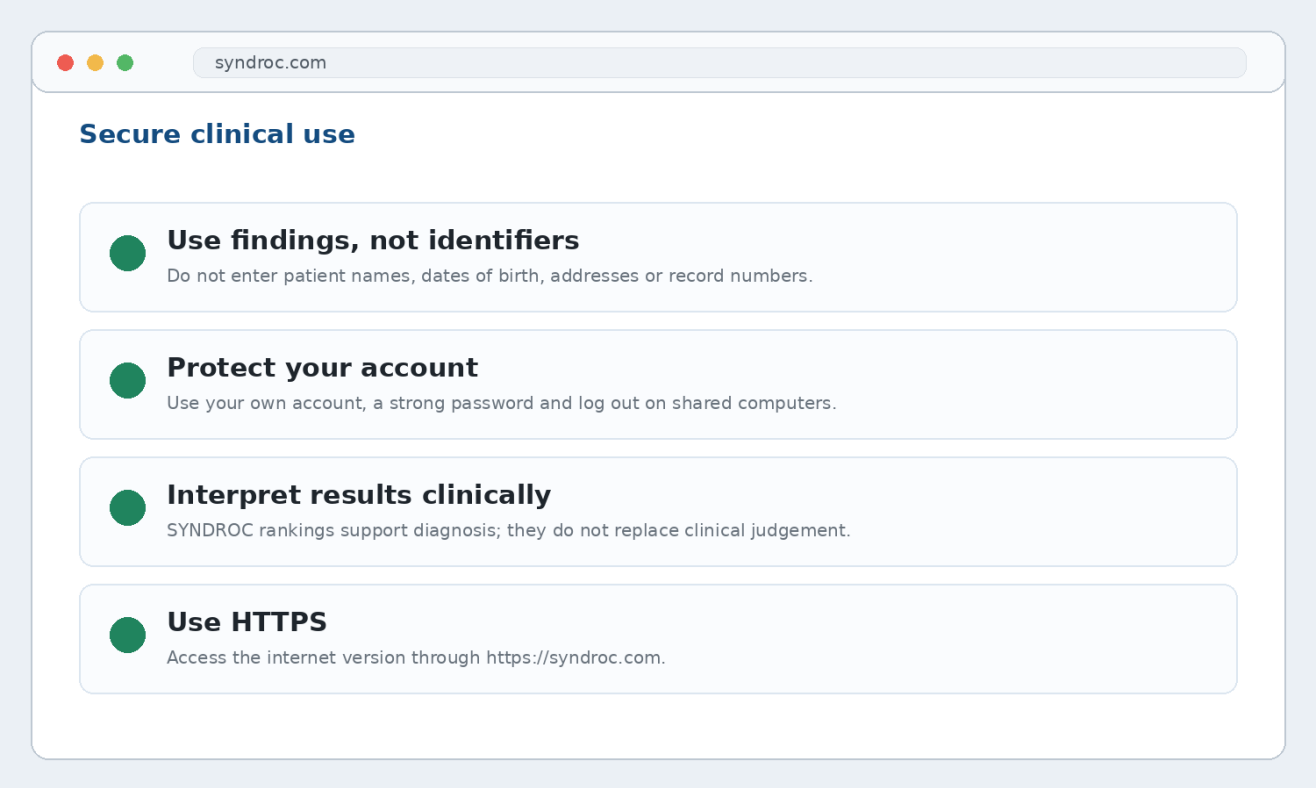


Figure 8 — Key rules for secure use of the internet version.

10.1 Do not enter patient identifiers

- Do not enter patient names.
- Do not enter full dates of birth.
- Do not enter addresses, telephone numbers or email addresses.
- Do not enter hospital or insurance identification numbers.
- Avoid free text containing information that could identify a patient.

10.2 Protect your user account

- Use only your own account.
- Keep your password confidential.
- Use Logout on shared computers.
- Do not leave an authenticated session unattended.
- Access SYNDROC through https://syndroc.com.

10.3 Clinical responsibility

SYNDROC supports clinical reasoning but does not replace history taking, examination, literature review, specialist judgement or confirmatory testing. The user remains responsible for the interpretation and clinical use of the result.

11. Troubleshooting

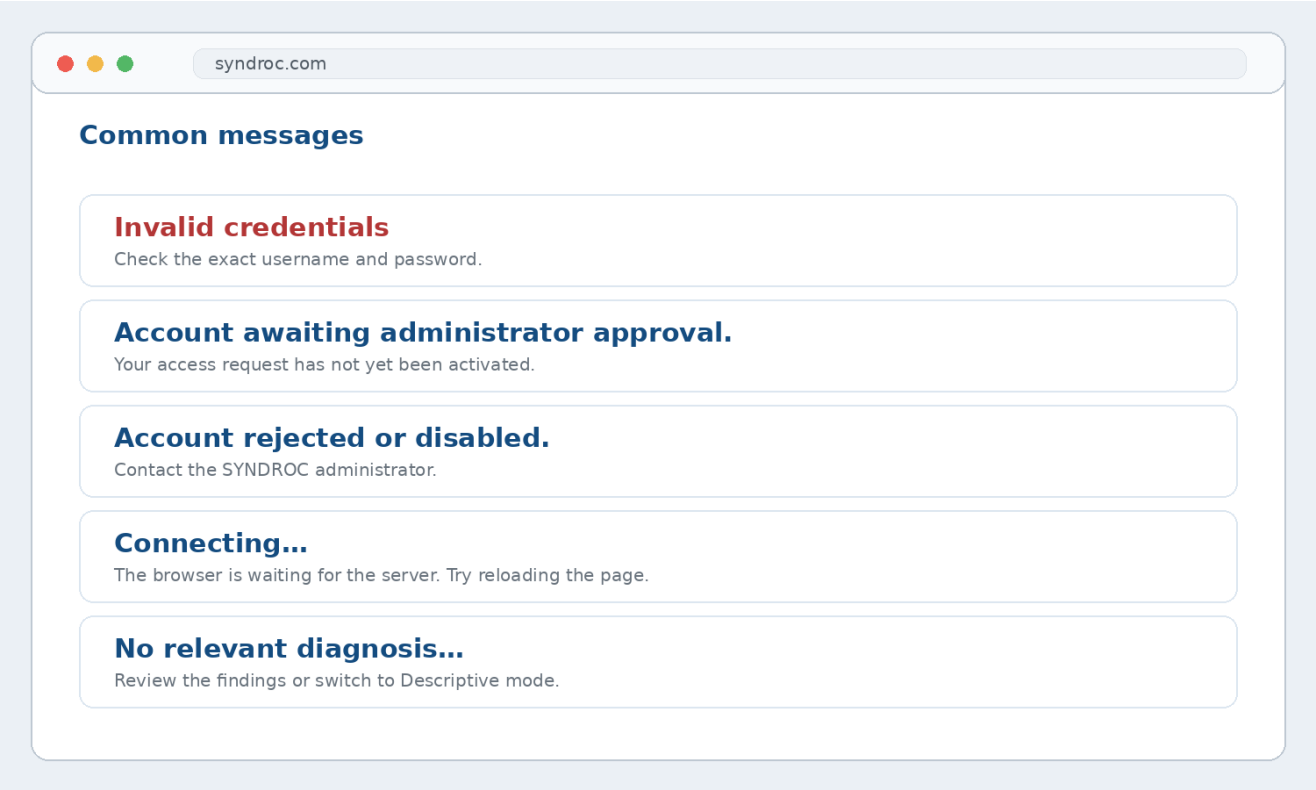


Figure 9 — Common user-facing messages and their usual meaning.

Problem	What to do
The login page does not open	Check the internet connection, reload syndroc.com, and try another modern browser if needed.
Invalid credentials	Check the exact username and password. Use Forgot password? if necessary.
Account awaiting administrator approval.	Wait for approval or contact the SYNDROC administrator if the request has been pending unusually long.
Account rejected or disabled.	Contact the SYNDROC administrator.
Connecting… does not finish	Reload the page. If the problem persists, try again later or report the problem to the administrator.
No relevant diagnosis…	Review the selected findings, remove uncertain findings, or try Descriptive mode.
No results to export	Generate a diagnosis before using Export Results.

11.1 Before reporting a problem

Please note the exact message displayed, the browser you are using, and the approximate time of the problem. A screenshot can be useful, but it should not contain patient-identifying information.

Appendix A — Keyboard shortcuts

Where	Key	Effect
Login page	Enter	Submit the form.
Suggestion list	↓ / ↑	Move through suggestions.
Suggestion list	Enter	Select the highlighted suggestion.
Suggestion list	Esc	Close suggestions.

Appendix B — Message reference

Message	Meaning
Invalid credentials	Wrong username or password.
Account awaiting administrator approval.	The account is not active yet.
Account rejected or disabled.	The account cannot sign in.
Connecting…	The browser is waiting for the SYNDROC server.
Failed to connect to SYNDROC server…	The server is unavailable or unreachable.
No relevant diagnosis…	Review the findings or try the Descriptive algorithm.
No results to export	Generate a diagnosis before exporting.

Appendix C — Quick reference card

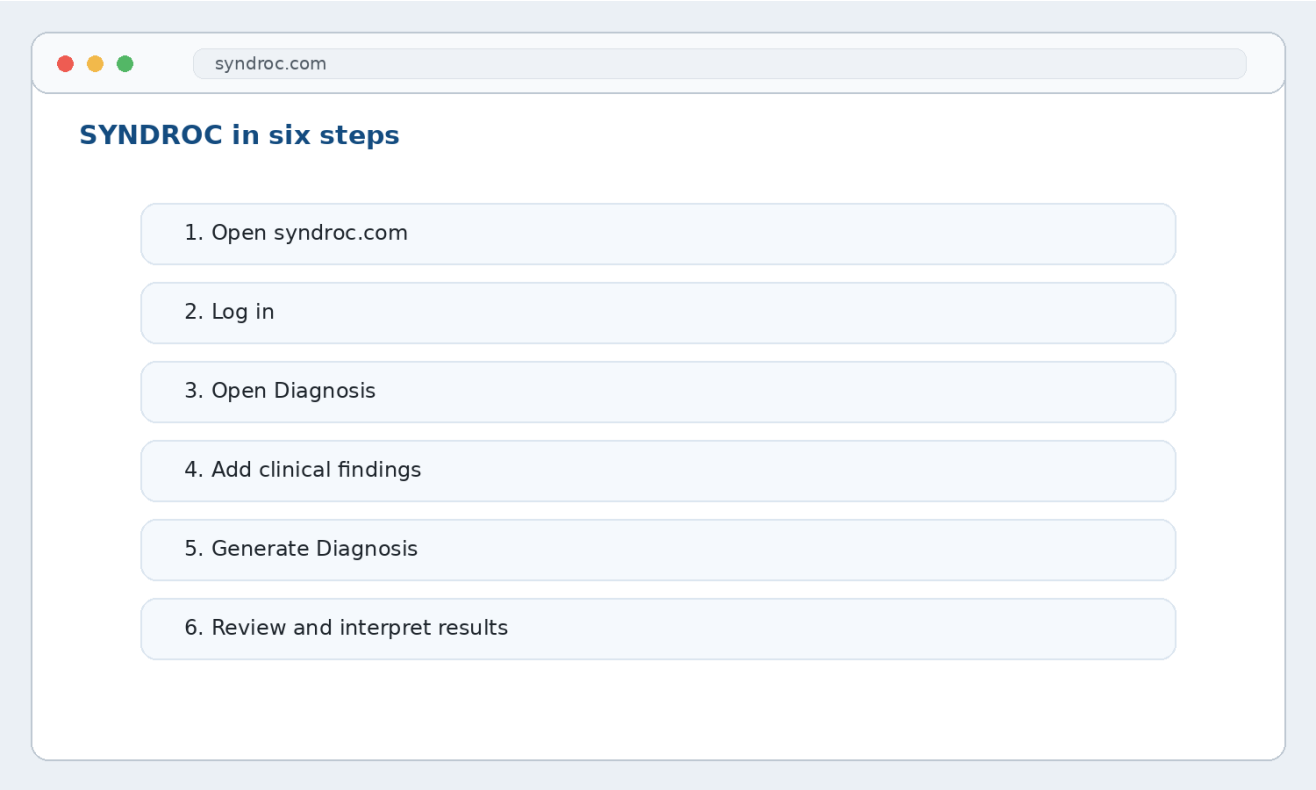


Figure 10 — Quick reference: the six-step user workflow.

To…	Do this
Open SYNDROC	Go to https://syndroc.com.
Request an account	Click Obtain access.
Reset a password	Click Forgot password? and follow the email verification process.
Add a finding	Type a few letters and choose a suggestion.
Run the search	Click Generate Diagnosis.
Broaden the differential	Switch to Descriptive.
Inspect a diagnosis	Open the diagnosis name in the result list.
Save results	Click Export Results.
Open teaching	Choose the Interactive Training Platform from the home page.
Log out	Click Logout.

About Dr Schorderet

Prof. hon. Daniel F. Schorderet is a medical geneticist and pediatrician whose clinical and academic work has focused on rare disorders, dysmorphology and the use of genetic information in diagnosis. He developed SYNDROC to help clinicians organize a patient's findings, consider possible syndromes and learn to recognize patterns of malformation.

After obtaining his FMH specialist qualification in paediatrics at the University of Geneva, he trained in genetics at the University of Washington in Seattle, at the Center for Inherited Diseases under Professors Stan M. Gartler and Arno Motulsky. On returning to Switzerland, he headed the Unit of Molecular Genetics in the Division of Medical Genetics at Lausanne University Hospital (CHUV) and later led the division. He subsequently became director of the Institute for Research in Ophthalmology (IRO) in Sion and was appointed professeur titulaire at the Health Sciences Center of the École polytechnique fédérale de Lausanne (EPFL). He retired in 2019 and now works as a medical geneticist in private practice. He has published more than 300 scientific papers.

The project began in the era of microcomputers. Its first publication appeared in 1984, when Dr Schorderet and Patrick Aebischer described SYNDROC in a letter to Archives of Disease in Childhood titled "Microcomputers and malformation syndromes." Their fuller research article, "SYNDROC: microcomputer based differential diagnosis of malformation patterns," followed in 1985.

Dr Schorderet has continued to develop SYNDROC as a clinical aid and teaching resource. The current internet version combines structured clinical findings, ranked differential diagnoses and interactive training. Its purpose is to support clinical reasoning and education; the final interpretation remains with the treating clinician.

He can be contacted at daniel.schorderet@unil.ch.

Selected early publications

Schorderet D, Aebischer P. Microcomputers and malformation syndromes. Archives of Disease in Childhood. 1984;59(10):1004. doi:10.1136/adc.59.10.1004-b.

Schorderet D, Aebischer P. SYNDROC: microcomputer based differential diagnosis of malformation patterns. Archives of Disease in Childhood. 1985;60(3):248–251. doi:10.1136/adc.60.3.248.