

pictures of standard syndromes and undiagnosed malformations



User Guide

For Windows and NT

POSSUM may only be used by or under the supervision of a qualified and duly licensed medical practitioner or medical scientist who is a geneticist dysmorphologist or paediatrician and who is an expert in the diagnosis of birth defects.

POSSUM functions as an electronic textbook about patterns of dysmorphism in humans and provides rapid access to information. However, POSSUM has been designed only to provide additional assistance to skilled practitioners in reaching diagnoses and for use as a teaching aid for persons who are not possessed of appropriate specialised or expert knowledge to provide appropriate training in birth defects. POSSUM does not make diagnoses and may not be used by any trainee for diagnostic purposes.

POSSUM contains information on many but not all syndromes and the information may be incomplete and is subject to future change. POSSUM may be disturbing to people unacquainted with the subject of dysmorphism.

Telemedia Software Laboratories and the persons who compiled the information and pictures in POSSUM, being The Murdoch Institute, the distributors of POSSUM and any corporation or institution which has installed POSSUM shall not be responsible for any decisions taken or inferences made by users of POSSUM.

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POSSUM 5.6 uses the POET Object Database.

POET is a registered trademark of POET Software Corporation

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The information in this document is subject to change without notice.

Contact Details:

We welcome any queries or suggestions that you might have. You can provide feedback via our web site: www.possu.net.au.

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1

Overview

Welcome to POSSUM, a tool that can help you to diagnose syndromes in your patients. This section includes the following topics:

- ◆ What is POSSUM?
- ◆ Background
- ◆ What's New in POSSUM?
- ◆ How We Maintain the POSSUM database
- ◆ Accessing the POSSUM Web Site
- ◆ References and Acknowledgments

What is POSSUM?

POSSUM is a software tool that can help you to diagnose syndromes in your patients and to learn about syndromes in general. POSSUM contains information on more than 2800 syndromes, including chromosomal and skeletal syndromes. The mediabase includes

- ◆ Pictures
- ◆ X-rays
- ◆ Diagrams
- ◆ Histopathology
- ◆ Video clips

By providing a flexible search facility, POSSUM saves you the time and effort usually involved in researching and cross-referencing syndrome information. You specify the traits exhibited by a patient and POSSUM provides a list of possible syndromes—including comments, citations, relevant pictures, sound and video. Of course, the final diagnosis continues to rely on your skills in visually recognizing and comparing features.

You can also use POSSUM to search for clinic patients that exhibit a set of traits or investigate the clinical patients associated with a syndrome.

Background

The POSSUM team was led by Professor David Danks and Dr. Agnes Bankier at the Murdoch Institute and the Victorian Clinical Genetics Services at the Royal Children's Hospital Melbourne. This team has used computer-aided syndrome identification since 1980.

The POSSUM system, with its first videodisc, was launched in March 1987 and in 1988 won the Australian Information Technology Award in the category of "Systems Innovation of the Year".

POSSUM is used by clinicians around the world and, given collaborative input, is helping to standardise syndrome terminology.

OSSUM

OSSUM contains information on skeletal dysplasia syndromes. In the past, OSSUM was a distinct product though its images were stored on the POSSUM videodisc. In POSSUM 5.0, the two products became one.

OSSUM was produced in collaboration with Dr. Hartmut Menger and Professor Jurgen Spranger from Mainz, Germany, together with Professor David Sillence, and Professor Kazimierz Kozlowski from Sydney, Australia, and Professor Pierre Maroteaux and Dr. Martine Le Merre from Paris.

What's New in POSSUM 5.6?

POSSUM's new 'Save As..' feature allows you to transport a POSSUM image for use within your lecturing material at the click of a button. This new feature saves any of the images within POSSUM as a bitmap image directly to a location you specify. Now you can easily include POSSUM in your PowerPoint™ presentations without cutting and pasting.

Improved image resolution – All of POSSUM images are now clearer. With higher resolution, enlarging the images using the 'Fit to window' feature in POSSUM results in a clearer image to assist with syndrome identification.

Animated POSSUM – while loading POSSUM, an animated POSSUM will now appear until POSSUM is fully loaded and ready to use.

Resolved issues running POSSUM on an Intel Pentium IV processor – This is a known problem with compatibility between Intel processors and a Java library file used by POSSUM. The updated library file is packaged with POSSUM 5.6, or POSSUM 5.5 users can simply contact POSSUM for the latest patch at support@possum.net.au.

POSSUM 5.6 includes the following additional features:

- An extended database containing over 2800 syndromes.
- 5000 new images, creating an image base of over 30,000 images —all available on CD-ROM.

How We Maintain the POSSUM Database

In the POSSUM database, each syndrome is described by a set of clinical or radiological traits—this information is drawn from a cross-section of the literature. The database is updated continuously as new publications about a syndrome become available.

To assess the traits that should be recorded for a syndrome we

- Refer to the journals in which the syndrome was reported
- Check the pictures that have been provided by the author
- Review the Online Mendelian Inheritance in Man (OMIM) entry where relevant

To maximise the probability of retrieval in a search, most traits reported in the literature are included in a syndrome description—regardless of how frequently they occur. If there is more than one way of describing a feature such as ‘the forehead’ several clinical and radiological traits may be included. A syndrome is an abstraction—individual patients do not manifest all the features of a syndrome.

Sometimes the description of a syndrome in the literature is vague or incomplete. For example, the available literature on a syndrome may state that “cardiac abnormalities” occurred in that syndrome. Where specific information about a trait is not available, we use the group trait so that it is not overlooked during a search. For more information about group traits, refer to Section 5. ‘Working with Traits’.

For many syndromes, we also maintain information about patients who have been diagnosed with that syndrome. These patients were seen in our clinic and photographed. Patient descriptions only include traits present in that patient.

We also maintain information about undiagnosed patients. If the traits exhibited by your patient match those of an undiagnosed patient, please contact us—we would be pleased to share more detailed clinical information about the patient, perhaps leading to the definition of a new syndrome.

Contributing to the POSSUM Imagebase

While many of our images are from the Royal Children’s Hospital in Melbourne, we invite authors of publications to contribute pictures to POSSUM.

If you submit pictures for POSSUM, your originals will be returned and you will be reimbursed for any replication or mailing charges. All contributors are explicitly acknowledged within the product.

Note: Material that has been previously published needs a release of copyright from the original publisher before it can be used in POSSUM. You need to let us know which of your illustrations have been published, and where, so that we can apply for release of copyright to use your pictures in POSSUM. Some journals insist that authors assign copyright on material and a subsequent release from the author is not necessary. Typically, copyright is jointly held, and releases from both the journal and the author are needed.

Accessing the POSSUM Website

You can access the POSSUM website while you are working with POSSUM (click POSSUM Home on the Toolbar). The website includes information about the latest POSSUM developments and a forum for your feedback. You can also subscribe to the POSSUM bulletin and receive regular email updates about the product.

To access the website you must have a web browser and an Internet connection.

References and Acknowledgments

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Boda D., Gyori I., Pasek B. (1989) Diagnostics of malformation syndromes using an available computer program, *Acta Paediatr. Hung.* 29:354-357

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2

Installing and Setting-Up

This section provides information about installing the POSSUM software. It includes the following topics:

- ◆ Software Requirements
- ◆ Hardware Requirements
- ◆ Installing POSSUM
- ◆ Defining Setup Options

Software Requirements

To run POSSUM you will need the following software:

- ◆ Microsoft Windows® 95 or 98 operating system or Microsoft® Windows NT® 4.0.
- ◆ An Internet web browser if you want to print POSSUM descriptions or visit the POSSUM website.
- ◆ The optional Internet features of POSSUM require Internet access—this access may require payment of a separate fee to a service provider.

Hardware Requirements

To run POSSUM you will need the following hardware:

- ◆ Personal or Multimedia computer with a 166 MHz Pentium microprocessor and at least 32 Mbytes of RAM
- ◆ Computer display with resolution of 1024 x 768 pixels and 256 colors (VGA).
- ◆ 304 Mbytes of available space on a hard disk drive. Pictures can be accessed directly from the POSSUM CD-ROM, or they can be copied to your hard disk drive and accessed from there. To copy the pictures to your hard drive you will need 954 Mbytes of free disk space.

Note: If required, the install can be split into 304Mbytes on one drive and 650Mbytes on another drive.

- ◆ CD-ROM drive.
- ◆ A mouse, or compatible pointing device.

- ◆ If you want to continue using a videodisc player to access images, you will need one of the following videodisc players:
 - Sony LDP (series 1500, 1550, 3600).
 - Pioneer LDV (series 2100, 2200, 2300, 2600, 4100, 4200, 4300, 4400).

The Pioneer LDV 4000 with IU-04 is no longer supported. Other players may be supported. Please contact us for more information.

Installing POSSUM

To install POSSUM:

1. Start Windows if it is not already running.
2. Insert the POSSUM CD in your CD-ROM drive.
 - a. Choose the Run option from your Windows Start menu.
 - b. Enter the letter for your CD-ROM drive followed by \possum—for example E:\possum.
 - c. Choose OK.
3. Read through the agreement and choose the I Accept button.
4. The Program Files dialog box is displayed. Unless you want to change the directory for the POSSUM installation, choose the Next button.

To change the destination of the POSSUM installation choose the Browse button and select the new location.

5. The POSSUM Images dialog box is displayed. You can choose to access images from the following locations:

Note: Each of these options is mutually exclusive. For example, if you decide after installation that you want to access images from your CD-ROM drive instead of the videodisc player, you will need to run POSSUM Setup again.

Option	Description
View Images from CD-ROM	<p>This option saves space on your hard drive by allowing you to access images from the POSSUM CD. This means that when you are using POSSUM, you will need to have the CD in your CD-ROM drive.</p> <p>POSSUM Setup automatically locates your CD-ROM drive, you can change this location by choosing the Browse button and selecting a new location.</p>
Copy all images to the hard drive	<p>This option copies all of the images from the POSSUM CD onto your hard drive. This means that you can work with POSSUM pictures independently of the CD. This option requires a minimum of 954 Mbytes of free space on your hard disk drive.</p> <p>When you choose this option, POSSUM Setup will automatically determine whether there is enough free space on your hard drive. If you do not have enough space on the currently selected drive, choose the Browse button to select another hard drive.</p>

Option	Description
View images from version 4.0 videodisc	<p>This option enables you to look at POSSUM pictures using your videodisc player.</p> <p>If you already use a videodisc player on this PC with POSSUM version 4.0 or 4.5, POSSUM Setup will automatically use the settings defined for the current videodisc player. If there is no videodisc player currently attached to your computer, you will be prompted to define the videodisc settings at the end of the Setup program.</p> <p>Note: Only images from POSSUM version 4.0 and 5.0 will be visible. Images for POSSUM 5.1 (or above) will not be available.</p> <p>To find out more about setting-up your videodisc player, refer to “Defining Setup Options” later in this section.</p> <p>For more information about the supported videodisc players refer to “Hardware Requirements” earlier in this section.</p>

6. Choose the Next button when you have chosen your image options.

If your computer has Internet access, the Internet Proxy Server dialog box is displayed. POSSUM Setup attempts to locate any proxy server information that exists for Internet Explorer© or Netscape Navigator©. If the fields in this dialog box are blank, it means that POSSUM Setup could not find the proxy information—this is probably because you do not access the Internet via a proxy server.

7. If you are unsure about this information, choose the Next button and define the details after you have installed POSSUM. For more information, refer to “Defining Setup Options” later in this section.
8. In the Registration Information dialog box, enter your details and choose the Next button. By registering your details we can make sure that you are kept up to date with the latest POSSUM developments. By default, your registration details will be automatically sent to us—if you do not want this to occur, uncheck the Send Registration Details via the Internet option.

You can also subscribe to the POSSUM bulletin which is a regular email outlining the latest POSSUM news.

These features are only available if you have an Internet connection.

9. The Install Checklist dialog box is displayed. If the list is correct, choose the Start Install button to begin the POSSUM installation. If you want to change any of your installation options choose the Back button

When installation is complete, you will be prompted to restart your computer.

To start POSSUM, choose the POSSUM 5.6 option from Programs under your Windows Start menu.

Removing POSSUM

To remove POSSUM from your computer:

1. Choose Settings from your Windows Start menu and choose the Control Panel option.

The Control Panel window is displayed.

2. Double-click the Add/Remove Programs icon.

The Add/Remove Programs Properties window is displayed.

3. Choose POSSUM 5.6 from the list.

4. Choose the Add/Remove button

The Select Uninstall Method dialog box is displayed.

5. Choose the Automatic option

6. Choose the Next button

7. Choose the Finish button.

All POSSUM files are removed from your computer.

Defining Setup Options

Once you have installed POSSUM you can define some Setup information using the Setup option from the POSSUM Toolbar.

Setting-Up Your Videodisc Player

The POSSUM Setup program automatically defines the settings for your videodisc player based on the player that is currently attached to your machine. If you do not have a videodisc player attached, you will be prompted to define the new settings at install time. When running POSSUM, you can change these settings at anytime. To change the setup information for your videodisc player:

1. Choose the Setup button from the POSSUM Toolbar.
2. Choose the Media button from the pop-up menu that is displayed.

The Media Settings dialog box is displayed.

Set the required options, using the following table for reference:

Option	Description
Video System	<p>In Australia, the UK and much of Europe, the 'native' video format is PAL.</p> <p>In the USA and Japan it is called NTSC.</p> <p>Generally, video monitors designed for PAL are incompatible with NTSC, though some video equipment is capable of coping with both formats.</p>
Player	<p>POSSUM 5.0 has been tested for use with the following video players:</p> <p>Sony LDP (series 1500, 1550, 3600)</p> <p>Pioneer LDV (series 2100, 2200, 2300, 2600, 4100, 4200, 4300, 4400).</p> <p>Note: The Pioneer LDV 4000 with IU-04 is no longer supported.</p>
Connect Using	<p>The serial communication port you are using to connect your computer to the videodisc player.</p>
Bits per second	<p>When you choose your player, this option is automatically set. You should check your videodisc player to ensure that it is the correct setting.</p>

Setting-Up Your Internet Proxy Information

Using POSSUM, you can link to OMIM over the Internet. To do this you need an Internet connection.

During POSSUM Setup, you are prompted to define information about your Internet proxy server. A network proxy is used to provide additional security between your computer and the Internet and to increase performance between networks by reducing redundant traffic. Not all Internet connections use a proxy server—for example, if you are accessing the Internet via a modem you will not need to define any proxy information.

If you are unsure about whether you use a proxy, you may need to ask your Internet administrator.

If you are using Internet Explorer or Netscape Navigator, POSSUM Setup will automatically use the proxy information that has been defined. When running POSSUM, you can edit the proxy information at anytime, to do this:

1. Choose the Setup button from the POSSUM Toolbar.
2. Choose the Network button from the pop-up menu that is displayed.

The Network Settings dialog box is displayed.

3. Define the following details:

Option	Description
Use a Proxy?	Uncheck this box if you do not use a proxy server.
Proxy Host	Enter the host name for the proxy server. You may need to ask your Internet administrator for this information.
Proxy Port	Enter the Port number for the proxy. You may need to ask your Internet administrator for this information.

4. Choose OK.

3

Quick Tour

Quick Tour provides an overview of the POSSUM workplace and helps you to become familiar with navigating around the system.

The POSSUM online help also provides animated demonstrations that show you how to work with POSSUM. Choose the Help button from the POSSUM Toolbar to access these demonstrations.

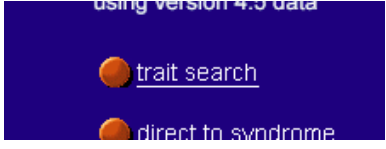
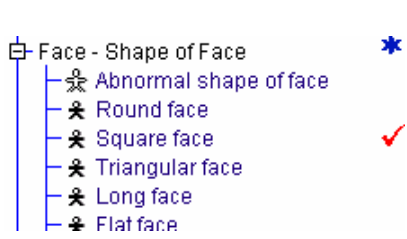
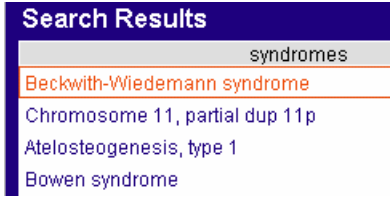
You can also run an online demonstration from the POSSUM web site (www.possu.net.au).

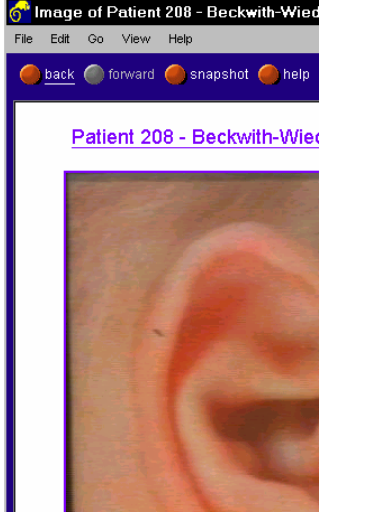
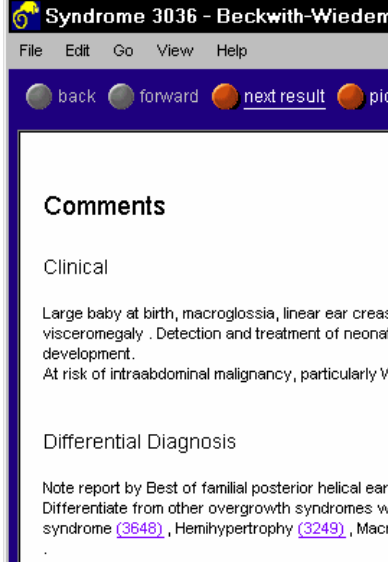
This section includes the following topics:

- ◆ Starting POSSUM
- ◆ POSSUM Basics
- ◆ POSSUM Setup
- ◆ Getting Help
- ◆ Closing POSSUM

POSSUM Basics

Most often, POSSUM is used to find syndromes based on the features or traits exhibited by a patient. The following table provides a high-level overview of this process using POSSUM:

0 Step	0 Illustration
<p>Start POSSUM and choose the Trait Search button.</p>	 <p>using version 4.3 data</p> <p>trait search</p> <p>direct to syndrome</p>
<p>From the Trait Selector, choose the most significant traits exhibited by your patient.</p> <p>As you go, look-up trait definitions in the Trait Atlas.</p> <p>Use search modes and the score threshold to refine your search. Refer to “Strategies for Selecting Traits” in Section 5.</p>	 <p>Face - Shape of Face *</p> <ul style="list-style-type: none"> Abnormal shape of face Round face Square face ✓ Triangular face Long face Flat face
<p>Run a syndrome search based on the traits you have selected.</p>	 <p>Search Results</p> <p>syndromes</p> <p>Beckwith-Wiedemann syndrome</p> <p>Chromosome 11, partial dup 11p</p> <p>Atelosteogenesis, type 1</p> <p>Bowen syndrome</p>

0 Step	0 Illustration
<p>1 Preview images associated with a candidate syndrome.</p> <p>2</p>	 <p>The screenshot shows a web browser window titled "Image of Patient 208 - Beckwith-Wiedema". The browser has a menu bar with "File", "Edit", "Go", "View", and "Help". Below the menu bar are navigation buttons: "back", "forward", "snapshot", and "help". The main content area displays a purple header "Patient 208 - Beckwith-Wiedema" above a close-up photograph of a human ear.</p>
<p>3</p> <p>4 Evaluate the candidate syndromes in the POSSUM browser:</p> <ul style="list-style-type: none"> ◆ Read the syndrome commentary and refer to the pictures that are available ◆ Compare your patient's features with those listed for the syndrome. ◆ Check the clinic patients who have been diagnosed with the syndrome ◆ If you have Internet access, click on the link to the syndrome reference in the Online Mendelian Inheritance in Man (OMIM) <p>5</p> <p>6</p>	 <p>The screenshot shows a web browser window titled "Syndrome 3036 - Beckwith-Wiedema". The browser has a menu bar with "File", "Edit", "Go", "View", and "Help". Below the menu bar are navigation buttons: "back", "forward", "next result", and "picture". The main content area displays a purple header "Syndrome 3036 - Beckwith-Wiedema" above a section titled "Comments". Under "Comments", there are two sub-sections: "Clinical" and "Differential Diagnosis".</p> <p>Clinical</p> <p>Large baby at birth, macroglossia, linear ear creases, visceromegaly. Detection and treatment of neonatal development. At risk of intraabdominal malignancy, particularly Wilms tumor.</p> <p>Differential Diagnosis</p> <p>Note report by Best of familial posterior helical ear pits. Differentiate from other overgrowth syndromes with syndrome (3648), Hemihypertrophy (3249), Macrosomia.</p>

Arranging Windows

If you are not familiar with working in a Windows environment, it may be useful to browse through the Windows help—choose Help from your Start menu to access it.

The POSSUM online help also provides an animated demonstration that shows you how to work with POSSUM windows.

The Windows Taskbar contains a button for each POSSUM window that is currently opened. You can bring a window into focus by choosing it from the Taskbar:



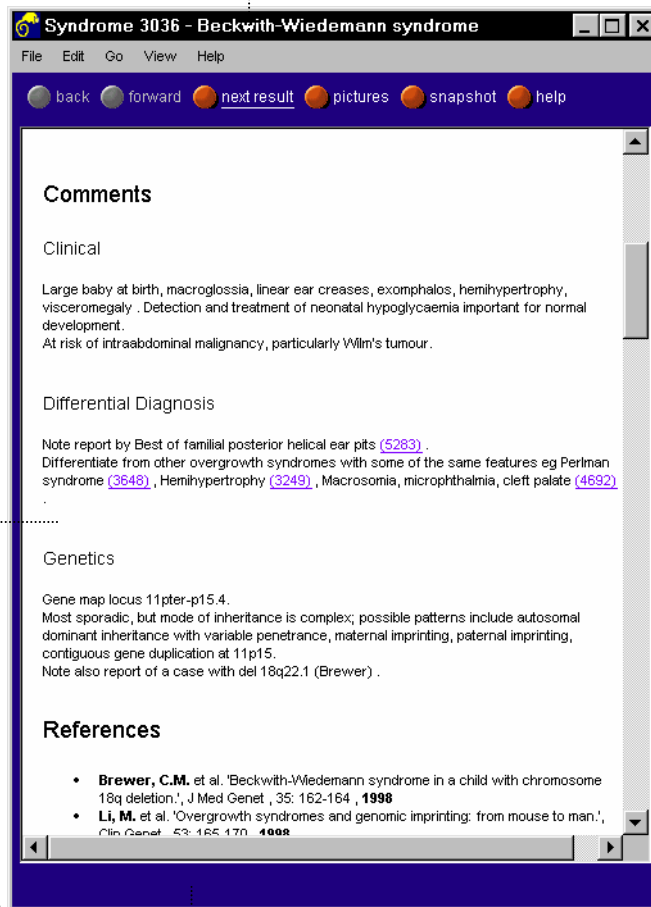
You can arrange and resize POSSUM windows to suit the way you want to work.

To move a window, click in the title bar and drag the window to the required position.

The controls in the top right corner of enable you to

- Minimize a window and place it on the Windows Taskbar
- Maximize a window and make it fill the whole screen
- Close a window

Click in the title bar and drag the window to the required location



Click and drag to make it wider

Click and drag to make it wider and longer

Click and drag to make it longer

Clearing Your Desktop

During a POSSUM session you may end-up with a number of windows opened on your desktop. For example, after running a trait search, you might have the following windows open:

- ◆ Trait Search window
- ◆ POSSUM Browser
- ◆ The Image Browser
- ◆ One or more ‘snap-shots’

To clean your desktop and get back to start-up mode, choose the Clear Desktop option from the POSSUM Toolbar. If the Toolbar is hidden below other windows, choose the POSSUM button from the Windows Taskbar to bring it into focus.

POSSUM Setup

When you install POSSUM you will be prompted to provide the required setup information. You can change this information using the Setup option from the POSSUM Toolbar.

Refer to the Section 2. “Installing and Setting-Up” for more information about the installation procedure and setup options.

Getting Help

While working with POSSUM you can access the online help at any time. If you choose the Help button from a specific window, you will get help that relates to that window. If you choose the Help button from the POSSUM Toolbar or from the Help menu, you will get a table of contents for all the available help topics.

The online information is displayed in the Help Browser. In this Browser you can move forward and backward through topics and follow links to find the information you need.

By positioning this window, you can follow instructions and interact with the application at the same time. Click on the title bar of this window and drag it to a convenient location on your desktop.




The online help contains a number of animated demonstrations that describe how to use POSSUM. You can find a list of these demonstrations by choosing the Help Topics button from the Help window.

Closing POSSUM

You must close a session from the POSSUM Toolbar. From the Toolbar you can

Click the Exit button.

OR

Click the  in the top-right corner of the Toolbar.

If the POSSUM Toolbar is obscured by other windows, choose the POSSUM button from the Windows Taskbar to bring it into focus

4

Searching with POSSUM

POSSUM can take some of the time and effort out of researching and cross-referencing syndrome information in the literature. Using the flexible searching facility, you can find syndromes or patients that match the traits exhibited by your patient. If required, you can bypass traits and go directly to a syndrome to find out more about it.

This section includes the following topics:

- ◆ Searching for Syndromes by Trait
- ◆ Searching for Clinic Patients by Trait
- ◆ Searching for a Specific Syndrome

Searching for Syndromes by Trait

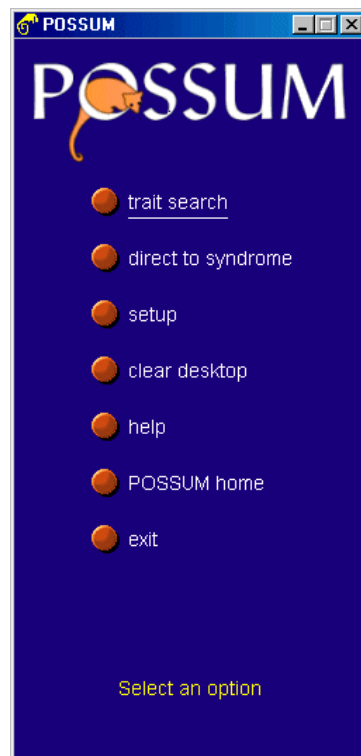
POSSUM can help you to find syndromes with features that resemble those of your patient. By comparing the description and pictures with your patient, you can get closer to a diagnosis.

Note: POSSUM is not an expert system or diagnostic tool, it is an aid to focusing on and evaluating the most likely syndromes.

To search for syndromes that match a set of traits:

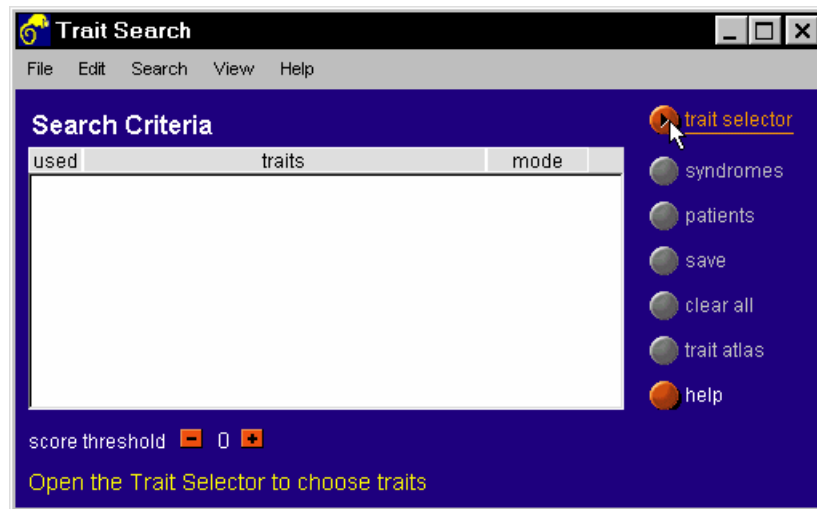
1. Start POSSUM by choosing it from your Windows Start menu.

The POSSUM Toolbar is displayed.



2. Choose the Trait Search button.

The Trait Search window is displayed.



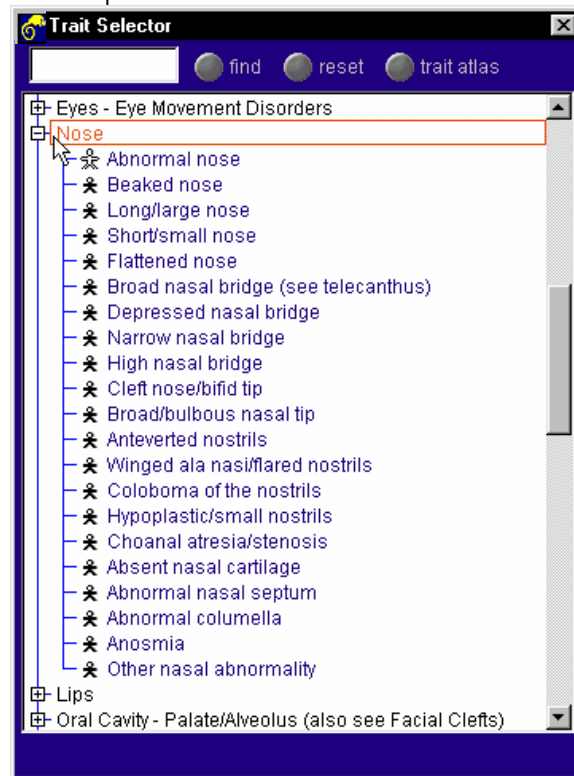
3. Choose the Trait Selector button.

The Trait Selector fold-out window is displayed.

4. Click on a category to display the traits associated with it. You can find a specific trait by entering the trait name in the Find field—refer to Section 5. 'Working with Traits' for more information.

To find a specific trait, enter the name (or part of it) and choose find

Click on Nose and all the traits relating to this category are displayed.



5. Click on a trait to add it to the Search Criteria.

A check mark is displayed beside the trait to indicate that it has been added. You can also see the trait displayed in the Search Criteria list.

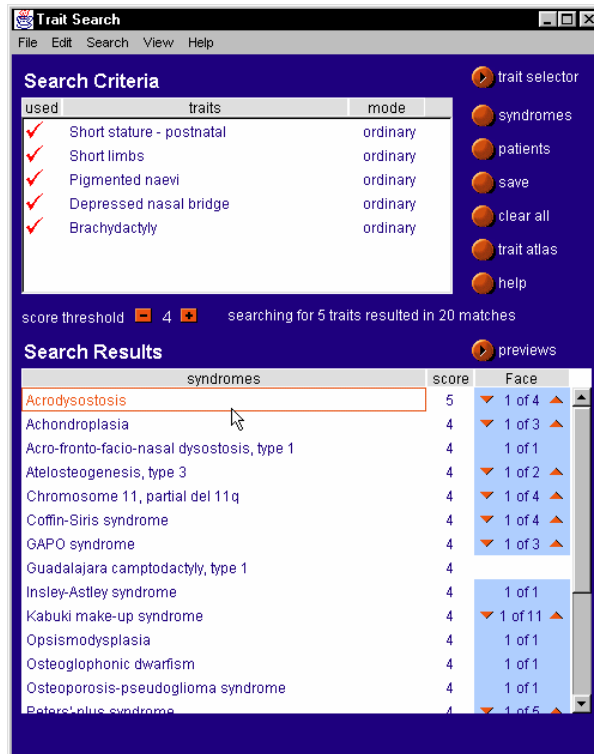
6. Add the significant traits that are exhibited by your patient.

Note: At this stage, it is best to choose only the most striking features. You can always come back and refine the Search Criteria later on. For information about selecting traits, refer to Section 5. 'Working with Traits'.

7. Close the Trait Selector fold-out window by clicking the Close Selector button.
8. Choose the Syndromes button.

The matching syndromes are displayed in the Search Results list.

The syndromes that match the selected traits are displayed in the lower half of the window.



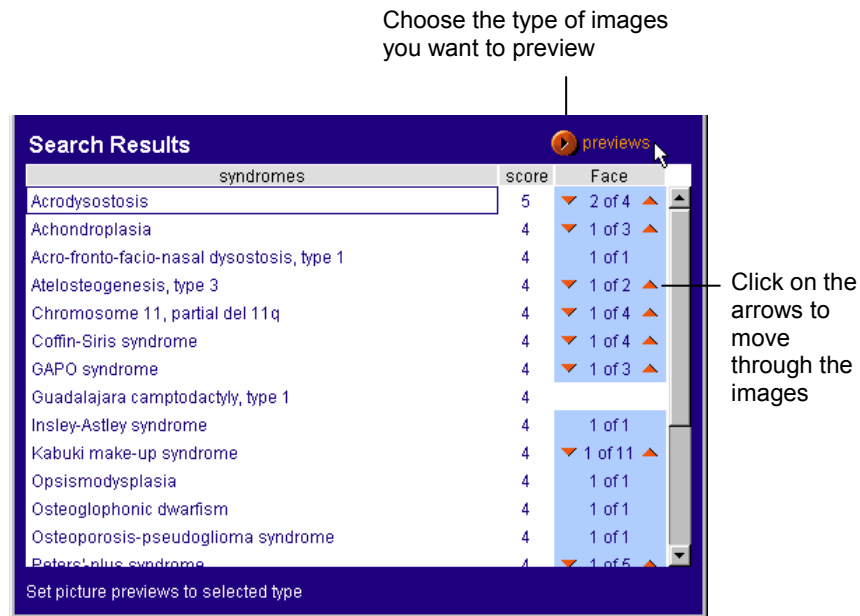
9. Click a syndrome name to display the syndrome description.

To find out more about working with the Search Results list and syndrome results in general, refer to Section 6. 'Working with Search Results'.

Previewing Images

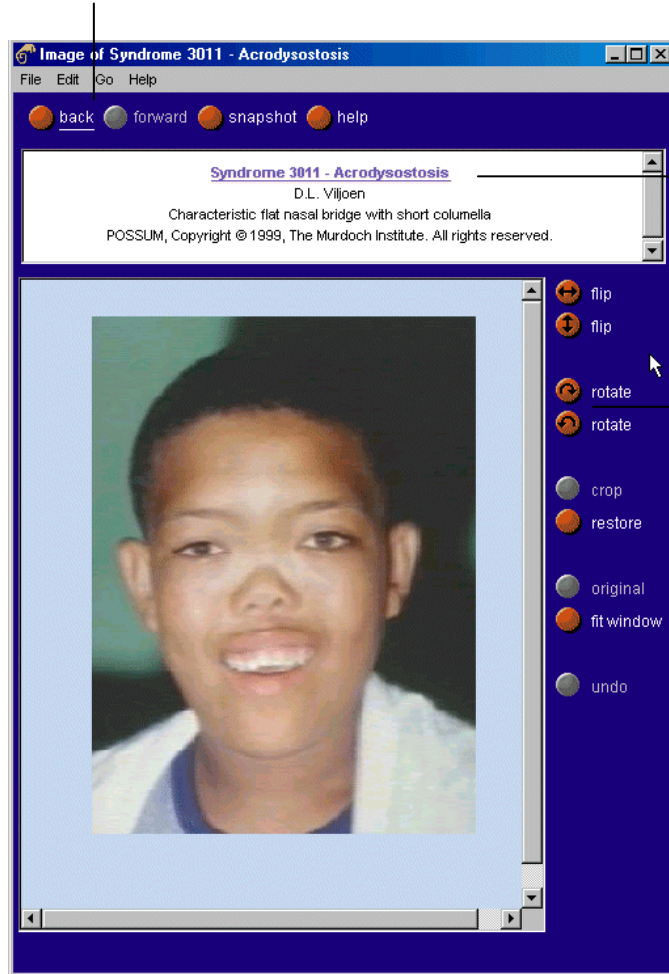
To preview the images that are available for a syndrome, click in the last column of the Search Results list and use the arrows to move backwards and forwards through the images. You can set the type of images you want to preview by selecting an option from the Previews drop-down list.

Note: If you are accessing images from the POSSUM CD-ROM, make sure Disc 2 is in the CD drive.



Images are displayed in the Image Browser. Each time you select an image it is added to the browse sequence in the browser. You can page backwards and forwards through selected images.

Use buttons to move backward and forward through selected images



Click on the syndrome title to see a text description of the syndrome

Use the buttons to flip, rotate or zoom-in on an image. For more information about manipulating images refer to Section 6. "Working with Search Results."

Searching for Clinic Patients by Trait

POSSUM contains information and pictures on patients seen in clinics at the Royal Children's Hospital in Melbourne, the Adelaide Children's Hospital, and some other centres that have contributed pictures to POSSUM.

Typically, you will search for candidate syndromes and then look at the associated clinic patients. If you are having trouble finding candidate syndromes, you might want to run a search on clinic patients instead. Perhaps your patient resembles another clinic patient who has not been diagnosed.

If you match your patient with an undiagnosed patient, the Murdoch Institute would be pleased to share with more detailed clinical information with you—perhaps leading to the definition of a new syndrome.

To search for patients by trait:

1. Choose the Trait Search button from the POSSUM Toolbar.
The Trait Search window is displayed.
2. Select the required traits from the Trait Selector fold-out window. For information about how to build your Search Criteria, refer to Section 5. 'Working with Traits'.
3. Choose the Patients button.
The matching clinic patients are displayed in the Search Results list.
4. Look at the previews available for a patient by clicking in the Preview column and using the arrows to cycle through the available pictures.

You can set the type of previews you want to look at by choosing the Previews button and selecting the type of preview you are interested in.

Note: If you are accessing images from the POSSUM CD-ROM make sure Disc 2 is in the CD drive.

5. Click the diagnosis to access the patient description in the POSSUM Browser.

For more information about evaluating clinic patients, refer to Section 6. 'Working with Search Results'.

Searching for a Specific Syndrome

Typically, you use POSSUM to search for syndromes based on the traits that are exhibited by your patient. You can also use POSSUM as a 'text book' by looking up specific syndromes to find out more about them.

Note: POSSUM syndromes are intended to optimise matching during trait searches and are not designed primarily for teaching about a syndrome. You should always consult the core literature for more detailed information, particularly where management decisions are being made.

You can search for a syndrome by

- ◆ Syndrome Name
- ◆ Author Name
- ◆ POSSUM ID
- ◆ OMIM Number

Searching by Syndrome Name

To search for a syndrome by syndrome name:

1. Choose the Direct to Syndrome option from the POSSUM Toolbar.

The Syndromes Search window is displayed with the Syndrome Name tab in focus:



2. Enter the name of the syndrome you want to find. You can also enter part of the name. POSSUM will also search for synonyms or common alternatives for a syndrome name.
3. Choose the Find button.

The matching syndromes are displayed in the syndrome list.

4. Look at the previews available for a syndrome by clicking in the Face column and using the arrows to cycle through the available pictures.

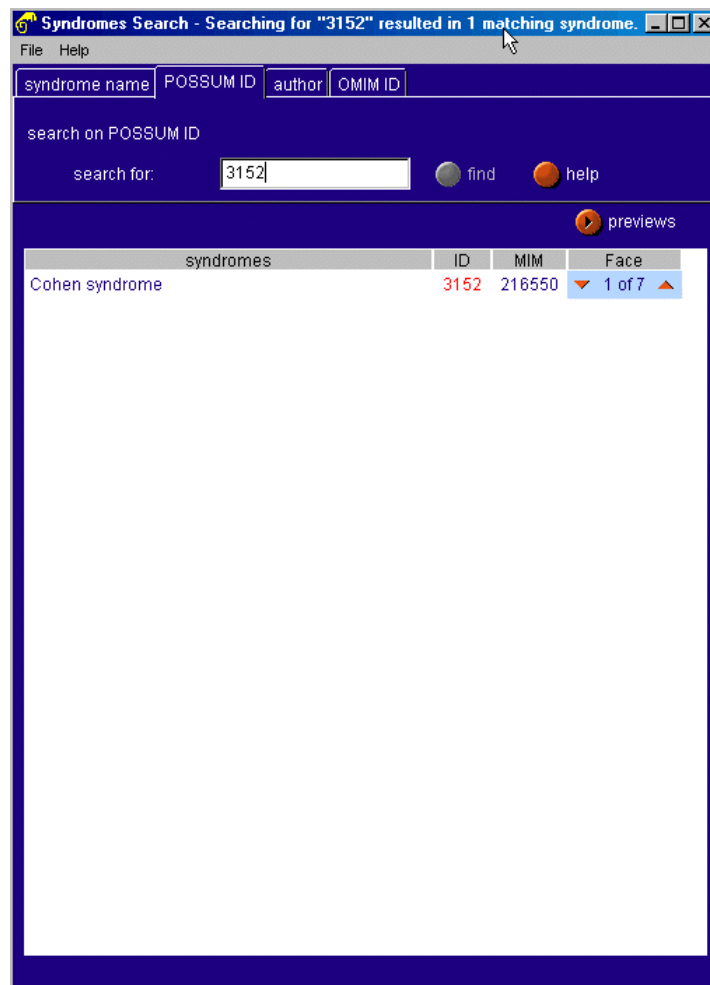
You can set the type of previews you want to look at by choosing the Previews button and selecting the type of preview you are interested in.

Note: If you are accessing images from the POSSUM CD-ROM, make sure the Disc 2 is in the CD drive.
5. Click a syndrome name to see its description displayed in the POSSUM browser.

Searching by POSSUM ID

Each syndrome in the POSSUM database is identified by a number. If you know the number, you can use it to find a syndrome. To search for syndrome by POSSUM ID

1. Choose the Direct to Syndrome option from the POSSUM Toolbar.
2. The Syndromes Search window is displayed with the Syndrome Name tab in focus.
3. Choose the POSSUM ID tab at the top of the window.



4. Enter the POSSUM ID in the Search For field.
5. Choose the Find button.

The matching syndromes are displayed in the syndrome list.

6. Look at the previews available for a syndrome by clicking in the Face column and using the arrows to cycle through the available pictures.

You can set the type of previews you want to look at by choosing the Previews button and selecting the type of preview you are interested in.

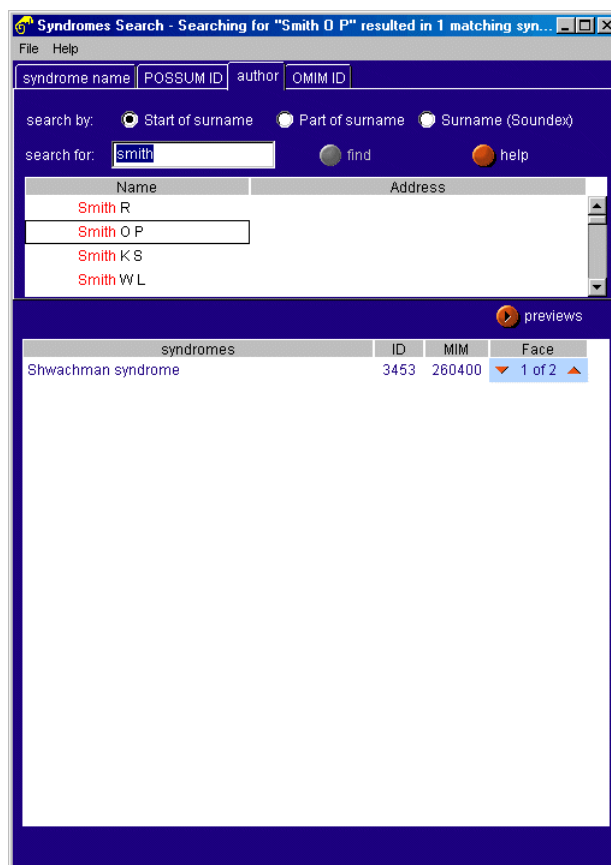
Note: If you are accessing images from the POSSUM CD-ROM, make sure the CD is in the CD drive.

7. Click a syndrome name to see its description displayed in the POSSUM browser.

Searching by Author Name

To search for syndrome by the name of the author who published it:

1. Choose the Direct to Syndrome option from the POSSUM Toolbar.
2. The Syndromes Search window is displayed with the Syndrome Name tab in focus.
3. Choose the Author tab at the top of the window.



4. Select an option from the Search By radio buttons:

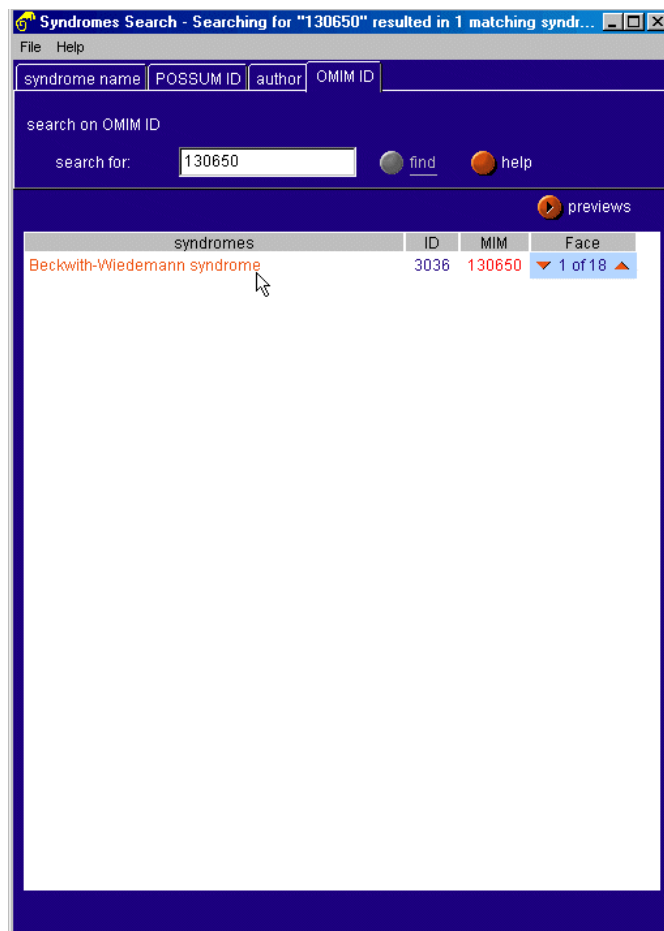
Option	Description
Start of Surname	Enter the letters at the start of the author name. POSSUM will find all the names that start with the specified letters.
Part of Surname	Enter any part of an author's surname. For example, you could enter 'man' and POSSUM will find all of the names that contain it such as "Amano" and 'Angelman'. This search takes slightly longer than the Start of Surname search.
Surname (soundex)	Enter a name that sounds like the one you are looking for. For example, if you enter 'Smith', POSSUM will find 'Schmid', 'Schmidt', 'Schmitt' and so on.

5. Enter the author name (or name part) and choose the Find button.
The Authors matching your search are displayed.
6. Click on the required author name.
The Syndromes published by the selected author are displayed.
7. Look at the previews available for a syndrome by clicking in the Face column and using the arrows to cycle through the available pictures.
You can set the type of previews you want to look at by choosing the Previews button and selecting the type of preview you are interested in.
Note: If you are accessing images from the POSSUM CD-ROM, make sure the CD is in the CD drive.
8. Click a syndrome name to see its description displayed in the POSSUM browser.

Searching by OMIM ID

Where possible, POSSUM provides an OMIM (Online Mendelian Inheritance in Man) reference number for the syndromes in the database. If you know the OMIM number, you can use it to find a syndrome.

1. Choose the Direct to Syndrome option from the POSSUM Toolbar.
2. The Syndromes Search window is displayed with the Syndrome Name tab in focus.
3. Choose the OMIM ID tab at the top of the window.



4. Enter the OMIM ID in the Search For field.
5. Choose the Find button.

The matching syndromes are displayed in the syndrome list.

6. Look at the previews available for a syndrome by clicking in the Face column and using the arrows to cycle through the available pictures.

You can set the type of previews you want to look at by choosing the Previews button and selecting the type of preview you are interested in.

Note: If you are accessing images from the POSSUM CD-ROM, make sure the CD is in the CD drive.

7. Click a syndrome name to see its description displayed in the POSSUM browser.

5

Working with Traits

POSSUM provides over 1100 traits that you can use to describe your patient. You need to think carefully about the traits you choose. Spend some time trying out different combinations of traits until you have a manageable list of likely diagnoses.

This section includes the following topics:

- ◆ Adding a Trait
- ◆ Removing a Trait
- ◆ Saving the Trait Search Criteria
- ◆ Setting the Search Mode for a Trait
- ◆ Finding a Specific Trait
- ◆ Finding Out More about a Trait
- ◆ Strategies for Selecting Traits
- ◆ Printing the Trait Dictionary

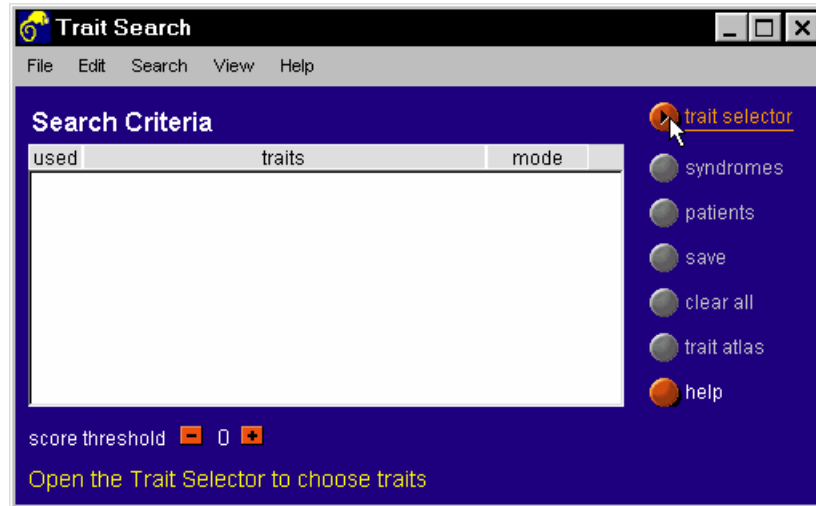
For overview information about running a trait search, refer to Section 4. “Searching with POSSUM”.

Adding a Trait

To add a trait to your Search Criteria:

1. Choose the Trait Search option from the POSSUM Toolbar.

The Trait Search window is displayed.

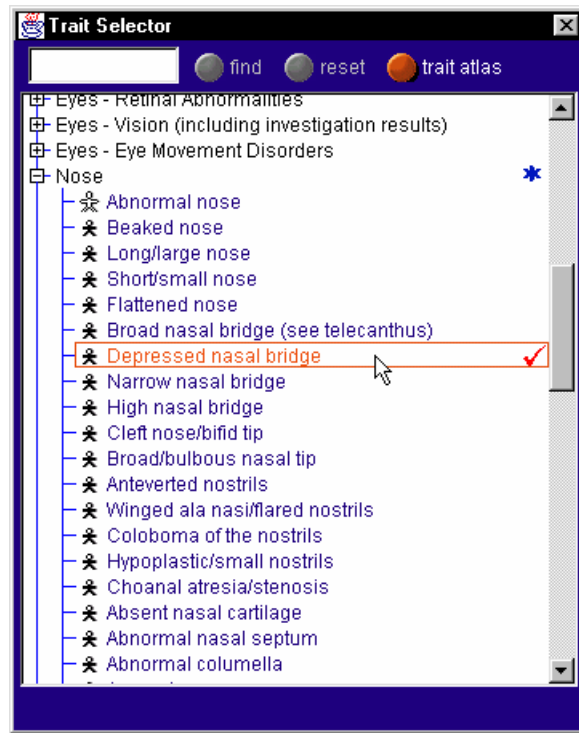


2. Choose the Trait Selector button.

The Trait Selector fold-out window is displayed.

3. Click on the category name that represents the part of the body you are trying to describe. The traits available under that category are displayed. To view the details and pictures for a trait, select it and choose the Trait Atlas button.

You can also use the find facility at the top of the window to locate a specific trait. Refer to “Finding a Specific Trait” later in this section.



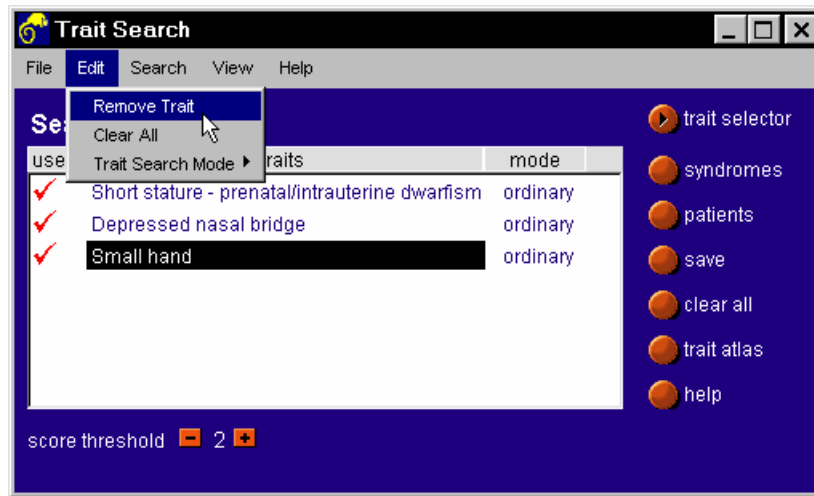
4. Click the required trait name.

The trait is added to the Search Criteria list. In the Trait Selector fold-out window, a checkmark is displayed beside the trait to indicate that it has been selected. An asterisk is displayed beside the category to indicate that a trait in that category has been selected.

Removing a Trait

To remove a trait from your Search Criteria:

1. In the Search Criteria, select the trait you want to remove.



2. Choose the Remove Trait option from the Edit menu.

OR

Click in the Used column for the trait you want to remove.

The trait is greyed and an ✖ is displayed to indicate that the trait will not be used in the search.

Removing all Traits

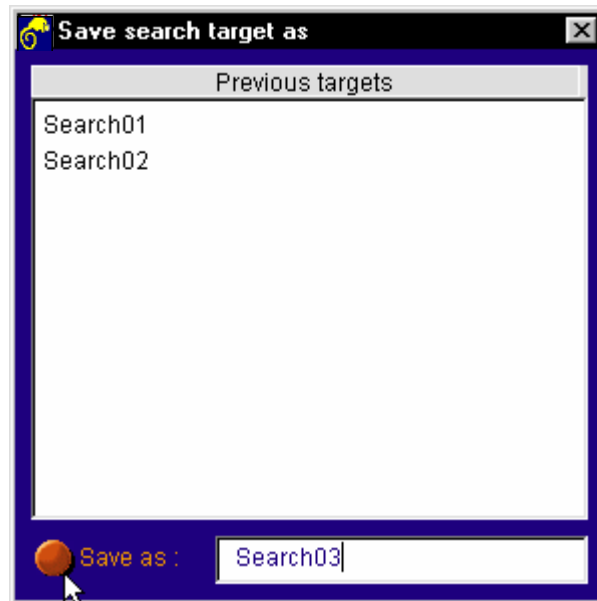
To remove all traits from the Search Criteria, choose the Clear All button in the Trait Search window.

Saving the Trait Search Criteria

You can save the list of traits that you have selected for a search. Once you save this list, you can open it and run the search at any time. To save the trait search list:

1. Select the required traits from the Trait Selector.
2. Choose the Save button.

The Save Search Target As dialog box is displayed.



3. Enter a name for the trait list in the Save As field.
4. Choose the Save As button.

The name is displayed in the title bar of the Trait Search window. You can add or remove traits if required. Choose the Save button to apply any updates.

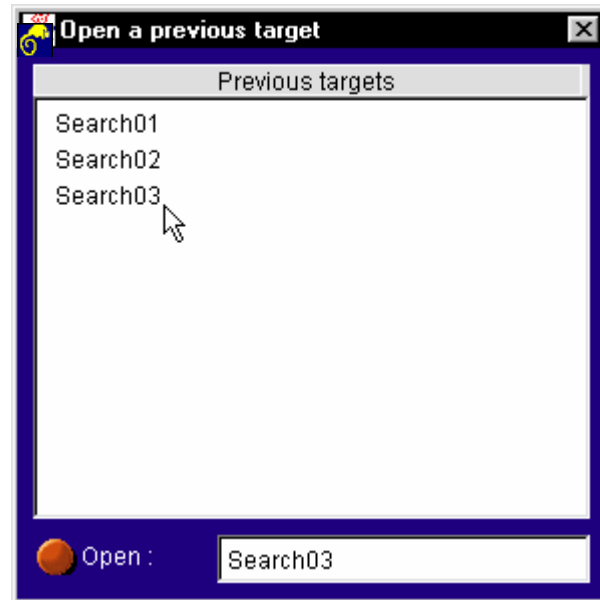
5. To start a completely new trait list, choose the Clear All button.

Opening an Existing Trait List

You can open a previously saved trait list in the Trait Search window. To do this

1. In the Trait Search window, choose Open from the File menu.

The Open a Previous Target dialog box is displayed.



2. Click on the trait list that you want to open.

The trait list is displayed in the Search Criteria panel.

You open add or remove traits from the list as required. To start a completely new trait list, choose the Clear All button.

Deleting a Saved Trait List

To delete a saved trait list:

1. In the Trait Search window, choose Delete from the File menu.
The Delete a Previous Target dialog box is displayed.
2. Click on the trait list that you want to delete.
The trait list is removed from the list of previous targets.

Setting the Search Mode for a Trait

As you build your Search Criteria, you can set a 'search mode' for each trait. Search modes can help you to refine your search so that the result list is more specific and easier to manage.

To change the search mode for a trait:

1. In the Search Criteria list, click in the Mode column for the trait you want to change.



used	traits	mode
✓	Short stature - postnatal	mandatory
✓	Short limbs	ordinary

2. As you click in the Mode column, the search mode changes.
The available modes include:

Mode	Description
<p>Ordinary</p>	<p>"Show me all the syndromes that have this trait."</p> <p>This is the typical mode that you use for searching. If you mark a trait as ordinary, each syndrome that has the trait will be displayed in the result list (this is subject to the effect of 'mandatory' and 'excluded' described below).</p> <p>This mode also finds any syndromes that are explicitly linked with the trait's group term.</p>
<p>Mandatory</p>	<p>"Don't show me a syndrome unless it has this trait."</p> <p>If your patient exhibits a single trait that is more striking than the others, you can mark the trait as 'mandatory'. If you make the trait mandatory, then all matching syndromes must contain it. If you are faced with an unmanageable list of matched syndromes, this provides a useful way of limiting your search.</p> <p>It is best to use the mandatory mode for one trait in your trait list. If you make multiple traits mandatory then the matched syndromes will be those that contain at least one of the mandatory traits. If you want all traits in the search list to be present in the matched syndromes then adjust the score threshold so that all traits must be matched.</p> <p>For more information about refining a search, refer to 'Strategies for Selecting Traits' later in this section.</p>

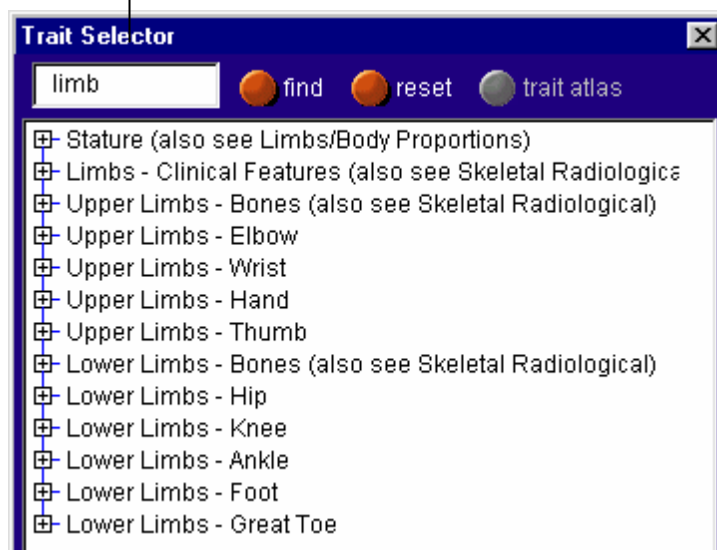
Mode	Description
Specific	<p>If you want to find a syndrome that is known to have been reported with a specific trait (usually rare) you can add the trait and set the search mode to specific.</p> <p>This mode is the same as ordinary mode except that the trait's group term is 'silenced' during the search. For example, a search on 'cardiac conduction defects/cardiac arrhythmia' in ordinary mode finds more cases (some with 'abnormal cardiovascular structure/function') than a search with the trait in specific mode.</p>
Excluded	<p>"Don't show me any syndromes that have this trait."</p> <p>If you set a trait to 'excluded' then syndromes with the trait will not be included in the result list. You should use with great caution. For example, you may decide to exclude the trait 'normal stature' when dealing with a seven year old patient who is obviously of short stature--this could lead to misleading results. Perhaps you are excluding a syndrome that exhibits normal stature at birth but changes as the patient ages. In this case, it would be better to make "short stature" a mandatory trait.</p> <p>If you set a group term to 'excluded' then members of the group are not automatically excluded. If you want to exclude a group of traits you have to set each of them to 'excluded'.</p>

Finding a Specific Trait

There are two ways to find a trait in the Trait Selector. The first way involves clicking on the category that best describes the region of the body you are interested in. You can then look at all the traits that are available in the category and find the best match. The second way involves searching for a specific trait by name, to do this:

1. In the Trait Search window choose the Trait Selector button.
2. Enter a name or part of a name in the Find field. For example, enter limb to find all the traits with limb in the title.

Enter a trait name or part of a name to find a specific trait



3. Choose the Find button.

The Trait Selector displays only the traits containing the specific text.

4. Click the trait that you want to add to the Search Criteria.
5. Choose the Reset button to re-display all of the available traits.

Finding out More about a Trait

As you build your Search Criteria, you may want to learn more about a specific trait. In POSSUM, all traits are contained in a Trait Atlas. The Trait Atlas enables you to

- ◆ Look at the images available for a trait.
- ◆ Read a trait description.
- ◆ Find out the trait population—how many syndromes and patients are linked to the trait.

To use the Trait Atlas

1. Click on a trait name in the Search Criteria list and choose the Trait Atlas button.

OR

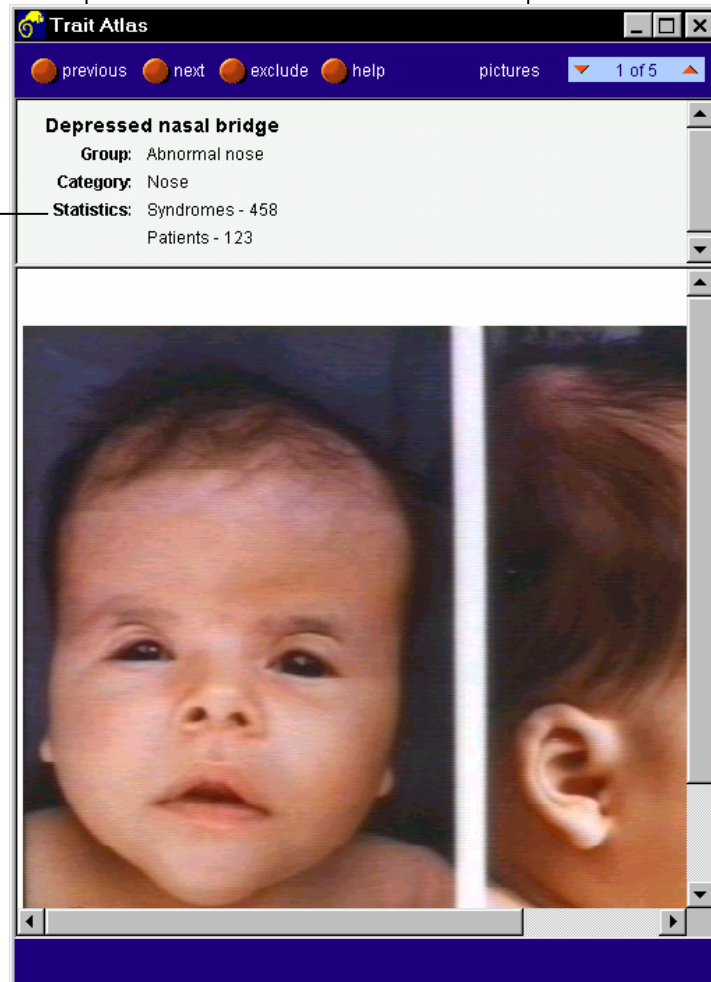
2. Click on a trait name in the fold-out Trait Selector and choose the Trait Atlas button at the top of the Trait Selector.

The Trait Atlas is displayed.

Choose the Next button to see the description for the next trait in the Trait Selector.

You can move through the pictures associated with the trait by clicking on the backward and forward arrows.

Statistics indicate whether the trait is common or rare



Strategies for Selecting Traits

In general, your searches should be based on the most striking features of your patient, not on a total description. You need to make decisions about which are the most significant traits. When searching for syndromes, you will get the best results if you use a small number of traits that are


- ◆ The most obvious and visually striking
- ◆ Do not occur frequently across a large number of syndromes

Check the Trait Atlas as you go, using the pictures to make comparisons with your patient.

Searching with POSSUM is fast and flexible, you can restructure and reorganize your Search Criteria until you are satisfied with the results.

Too Many Matching Syndromes?

If you run a search and find that there are too many matching syndromes:

1. Review the traits that are in your search criteria:
 - ◆ Is this the best and most concise description of your patient?
 - ◆ Have you used uncommon traits where possible? If you use traits that are common to many syndromes, you increase the number of matching syndromes. You can find out if a trait is common by checking its statistics in the Trait Atlas. If your patient exhibits a trait that is quite rare, try including only that trait.
 - ◆ Are your traits too general? Where possible try and choose specific traits rather than group traits. Group traits make your search more general because each trait in the group is included in the search. In the Trait Selector, group traits are represented by the symbol .

2. If you have revised your trait selections and still you are finding too many matching syndromes, you can try adjusting the score threshold.

The score threshold determines how many traits should match before a syndrome is considered a candidate. For example—if the threshold is 4, each matching syndrome must contain at least 4 of the traits. This helps you to zero-in on the most likely syndromes—the higher the score threshold, the more limited the search results. You should be careful about setting the score threshold too high, as you might exclude possible diagnoses. To adjust the score threshold, use the plus and minus buttons on the Trait Search window:



3. Another way of refining your search is to mark a single trait as mandatory.

Making a trait mandatory indicates that you are not interested in a syndrome unless it contains this trait. You can change a trait to mandatory by clicking in the Mode column. Keep clicking in the Mode column to cycle through the available search modes.



used	traits	mode
✓	Short stature - postnatal	mandatory
✓	Short limbs	ordinary

For more information about search modes, refer to “Setting the Search Mode for a Trait” earlier in this section.

4. If you have tried the options described above and are still left with an unmanageable set of search results, you can try using the Excluded search mode.

For example, if you are dealing with a 10 year old patient, you may not be interested in any syndromes that include the trait "Stillbirth/neonatal death". To indicate this, you add the trait to your search criteria and then mark it as excluded. You should use this option with caution, to avoid overlooking possible diagnoses—for example, the syndrome you are looking for may sometimes result in "Stillbirth/neonatal death".


No Matching Syndromes?

If you run a search and do not get any matching syndromes:

1. Try decreasing the score threshold.

The score threshold is automatically set to about half of the number of traits you have selected—if you have four traits, the threshold is set to two. If you are not getting any matching syndromes with the current score threshold, decrease it and run the search again.

2. Maybe some traits in your search criteria are too specific, try using group traits.

To broaden the scope of your search, replace a specific trait with the trait's group term. In the Trait Selector, group traits are represented by the symbol .

3. If you are having trouble finding syndromes that match your selected traits, try searching for clinic patients.

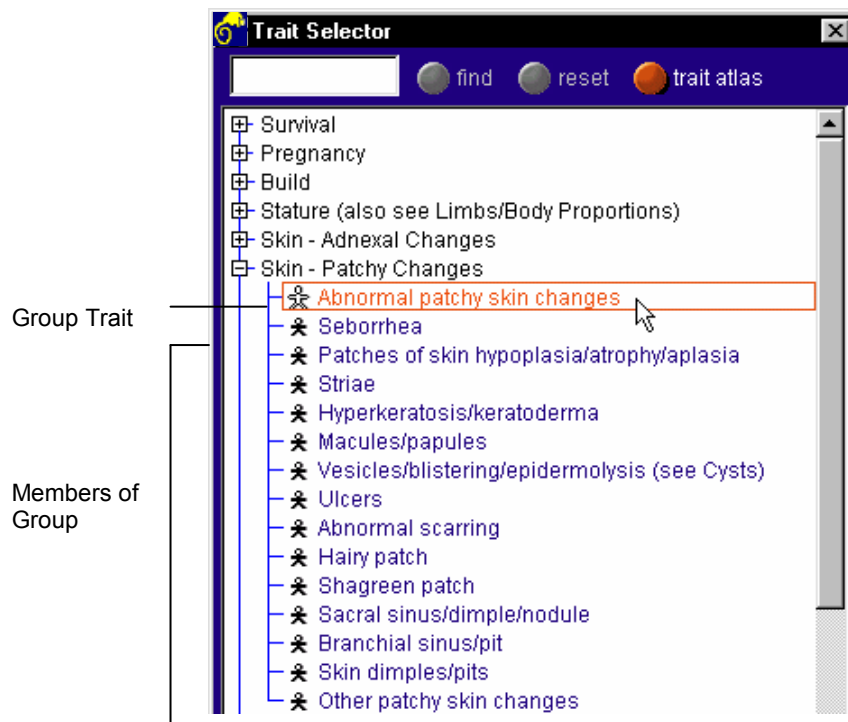
POSSUM contains information and pictures on patients seen in clinics at the Royal Children's Hospital in Melbourne, the Adelaide Children's Hospital, and some other centres that have contributed pictures to POSSUM. Perhaps your patient resembles another clinic patient who has not been diagnosed.

4. Choose the most significant and uncommon feature exhibited by your patient, search on that trait alone.

Working with Unknown or Ambiguous Traits

It is not always possible for you to supply the precise traits to describe a syndrome or a patient. For example, you may know that a patient exhibits abnormal skin patches but have no further information about the type of abnormality (seborrhea, striae, dimples, etc.).

To cater for this situation, POSSUM provides 'group traits' within each trait category. If you are not sure about specific traits, you can use the group trait. Group traits usually begin with the word 'abnormal'.



Group traits encompass all the traits in the group. For example, searching for 'Abnormal Patchy Skin Changes' will give you the same results as a searching for all the individual traits in the group. You should choose specific traits where you can. Using group traits makes your search less specific and increases your chances of obtaining results that do not resemble your patient.

Note: A group trait can only contribute a count of one to a search result's score, even though it implicitly includes all the traits that belong to the group.

Can't Find the Appropriate Trait?

If you can't find a suitable trait in the Trait Selector, you should use the group trait that is the closest match.

Although most trait categories include the trait 'other', this is intended for use by the developers of POSSUM rather than as a trait to include in your search criteria.

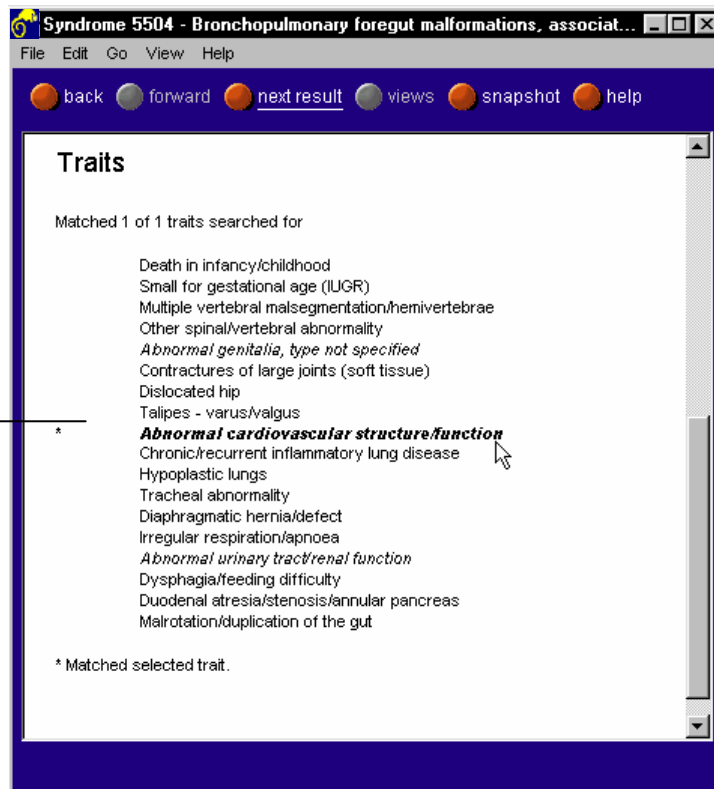
Syndromes that are Linked to a Group Trait

Sometimes a syndrome may be directly associated with a group trait. For example, the available literature on a syndrome may state that 'cardiac abnormalities' occurred in that syndrome. In the absence of further details, the syndrome will be directly associated with the trait 'Abnormal cardiovascular structure/function'.

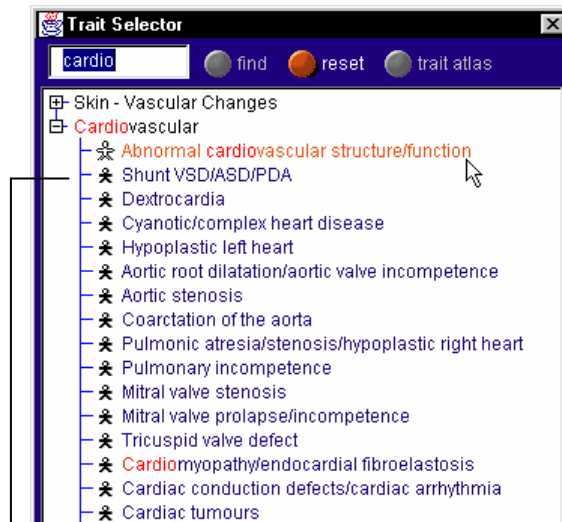
This tends to occur with chromosomal syndromes when the specifics are not readily available, or where a variety of cardiac defects may occur in patients with the same syndrome. Associating a syndrome with a group trait means implicitly that the syndrome manifests one or more traits in the group, but the specific trait is unknown or ambiguous. If you search for a specific trait within the group, POSSUM also checks for cases coded with the group term, since such cases might manifest the trait you are seeking.

For example, if you search for the specific trait 'cardiac conduction defects/cardiac arrhythmia', any syndrome explicitly associated with the group trait 'abnormal cardiovascular structure/function', will be included in the list of candidate diagnoses. You will need to evaluate cases associated with the group trait on other criteria, because the specific cardiac conduction defect may not be part of that syndrome. Alternatively, you can constrain the search to cases specifically associated with a conduction defect by using the specific mode for 'cardiac conduction defects/cardiac arrhythmia'.

This syndrome is linked to a group trait...



So...searching with any of the traits in the group will find the syndrome



6

Working with Search Results

When you have refined your trait search and have a manageable list of candidate syndromes, you can begin to look more closely at the search results. This section includes the following topics:

- ◆ Working with the POSSUM Browser
- ◆ Looking at Pictures
- ◆ Evaluating Syndromes
- ◆ Looking at Clinic Patients
- ◆ Printing Syndrome and Patient Descriptions

Working with the POSSUM Browser

When you have obtained a list of Search Results, you can use the POSSUM Browser to look more closely at each result.

0

1

2 Click a result to view it in the POSSUM Browser.



For information about performing a search, refer to Section 4. 'Searching with POSSUM'.

The POSSUM Browser is displayed containing information about the selected result.

- 0 Click on the MIM number to open the POSSUM web browser and go to the Online Mendelian Inheritance in Man (OMIM)

0

1

2

3

4

5

6

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8

9

10

11

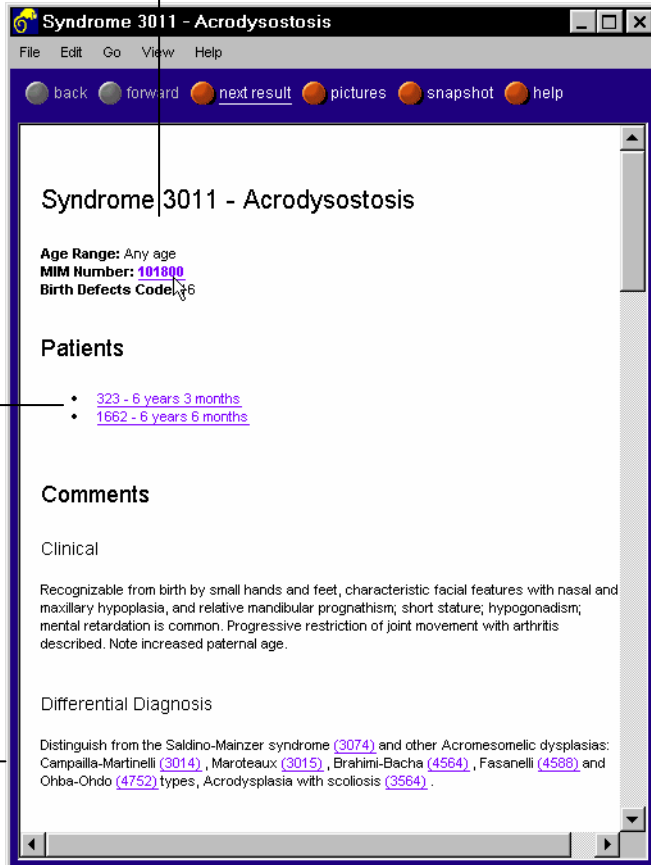
12

13

14

Click on a patient to see the description

Click on other syndrome numbers in order to make comparisons



0

To look at the next result in the Search Result list, choose the Next Result button—or you can go back to the Search Result list and click another result.

As you investigate each result, it is added to the ‘browse sequence’—like pages in a book. You can use the forward and back buttons to move between these ‘pages’.

Looking at Pictures

While you have a syndrome or patient description displayed in the POSSUM Browser, you can look at the pictures that are available. These might include:

- ◆ Photographs
- ◆ X-rays
- ◆ Diagrams
- ◆ Histopathology
- ◆ Video Clips

Note: If you are accessing images from the POSSUM CD-ROM, make sure Disc 2 is in the CD drive.

To look at pictures:

1. Choose the Pictures button from the POSSUM Browser toolbar.

The Picture Selector is displayed at the bottom of the window. To hide the Picture Selector, click on the Pictures button again.

0 Click on an arrow to move through the images

0 Click on a patient to display the description

0 Drag this scroll bar to see all the available picture types

Age	Hand	Head	Face	Ear	FrontView
Syndrome					
Any age	▼ 1 of 2 ▲	▼ 1 of 8 ▲	▼ 1 of 3 ▲		
0-2 years					
2-6 years					
6-16 years					
Patient					
6 years 3 months	1 of 1	1 of 1			1 of 1
6 years 6 months	1 of 1	1 of 1	1 of 1	1 of 1	1 of 1

2. Click in the cell that matches the age range and body part you are interested in. Syndromes are displayed in the upper half of the matrix—patients are displayed in the lower half.
3. Use the arrows to move forward and backward through the available pictures.

The Image Browser is displayed containing the selected picture.

Each time you select a picture from the Picture Selector it is displayed as a page in the Image Browser. Using the Back and Forward buttons, you can move through the selected images.

0 Use the Back and Forward buttons to move through the pictures you have selected

1

2

Syndrome 3088

3

4

Click the Snapshot button to keep a copy of the image on your desktop

Use these buttons to flip or rotate an image.

Use these buttons to crop and resize an image.

Use these buttons to zoom in and out of an image.

Use this button to save the image as an external file.


Manipulating Pictures


Using the buttons down the right-hand side of the Image Browser, you can manipulate the image that is displayed.

Changes to an image are only temporary, they are not saved to the POSSUM mediabase. When you close the Image Browser or look at another image, the changes that you made are lost. To keep the

changes on your desktop (while you look at other images), you can take a snapshot. For more information, refer to “Taking a Snapshot” later in this section.


Flipping an Image


To flip an image horizontally, choose the  button. To return the image to its original state, click the button again.

To flip an image vertically, choose the  button. To return the image to its original state, click the button again.

You can undo any changes that you make to an image by choosing the Undo button.

Rotating an Image

To rotate an image anti-clockwise, choose the  button. The image is rotated by 90 degrees each time you click the button.

To rotate an image clockwise, choose the  button. The image is rotated by 90 degrees each time you click the button.

You can undo any changes that you make to an image by choosing the Undo button.

Cropping an Image

To look more closely at part of an image:

1. Click in the image and drag a square around the area that you are interested in.
2. Choose the Crop button.

Only the cropped area is displayed in the Image Browser.

3. To make the cropped area fill the window, choose the Fit Window button.
4. To restore a cropped image to its original state, choose the Restore button.

Note: Changes to an image are only temporary, they are not saved to the POSSUM mediabase. When you close the Image Browser or look at another image, the changes that you made are lost. To keep the changes on your desktop (while you look at other images), you can take a snapshot. For more information, refer to “Taking a Snapshot” later in this section.

Making an Image Fit the Window

To resize an image so that it fills the Image Browser window, choose the Fit Window button.

This is useful if you want to

- ◆ avoid using scroll bars to see all of a large image
- ◆ enlarge an area that you cropped using the Crop button

To restore an image to its original size, choose the Restore button.

Undoing Changes to an Image

By clicking the Undo button, you can undo the last change that you made to an image. Since POSSUM keeps a list of all the changes you have made, you can keep clicking Undo until you are back to where you started.

Taking a Snapshot

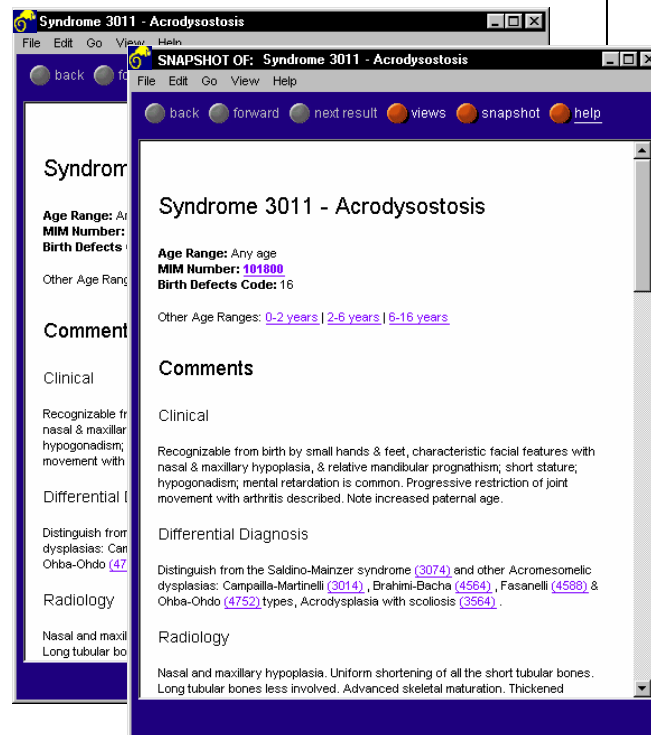
When investigating a syndrome or patient, you may want to keep related data or images on your desktop while you look at other syndromes or patients. This is a useful way to perform side-by-side comparisons of pictures and case descriptions.

Since each case description is displayed in the POSSUM Browser like a page in a book, you can page backward and forwards to compare descriptions. An alternative way of making comparisons is to take snapshots (make copies) of windows for side-by-side comparison on your desktop.

To keep a case description or image on your desktop, choose the Snapshot button from the POSSUM Browser or the Image Browser. You cannot create a snapshot from a snapshot.

You can arrange your windows to compare them or you can minimize the snapshot to the Windows Taskbar so that you can refer to it later on.

- 0 Click the minus sign to minimize a snapshot to your Windows Taskbar.



Saving an Image

When preparing lecturing material using POSSUM, images can be placed directly into your presentation media by selecting the new 'Save Image' Button on the Image Browser. This is a useful way to use the pictures to assist in teaching about the syndromes.

To use the 'Save Image' feature, have the image you require open in the Image Browser. Click on the 'Save As..' button located at the top of the Image Browser. A window will open allowing you to select the location that you wish to save the image file to. The image can be saved in bitmap, gif or zzz format without any cutting and pasting required. There are privacy requirements to restrict the access of images to licensed medical practitioners, health professionals and

students for clinical and teaching purposes. The images also have related copyright agreements and any use of and distribution of the images must not infringe these agreements.

Evaluating Syndromes

POSSUM is not an expert system or a diagnostic tool. While it can help you to focus on the possible diagnoses, you must still draw on your experience and expertise when evaluating a syndrome. When examining a syndrome description it might be useful to:

- ◆ Compare your patient’s traits with those listed in the syndrome description and those illustrated in the pictures. If your patient does not exhibit a trait that is listed for a syndrome you may want to go back and check your patient. For example, the syndrome may include radiological traits that you have not yet investigated.
- ◆ Check the clinic patients who have been diagnosed with the candidate syndrome. Looking at case studies may give you a better idea of the range of traits that are exhibited by patients with the syndrome—it is very rare for a patient to display all the traits that have been recorded for a syndrome.
- ◆ Research the articles that are listed in the syndrome description.
- ◆ Check the syndrome commentaries to see if the syndrome has been compared with or distinguished from other syndromes.

Looking at Syndrome Descriptions

A syndrome description includes the following information:

Label	Description
Age Range	<p>Displays the age range for the current description. Where a syndrome is only represented at one age, this is displayed as ‘Any Age’</p> <p>For some syndromes that are progressive with age, POSSUM provides separate descriptions. The following age ranges are possible:</p>

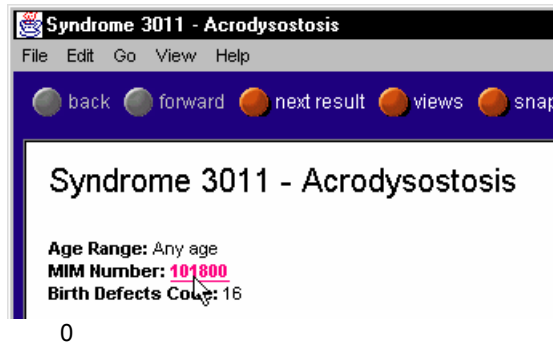
	Baby Toddler Child Adolescent Adult
MIM Number	Each syndrome in POSSUM is cross-referenced to <i>McKusick's Mendelian Inheritance in Man</i> . Click on the MIM number to access the syndrome in the online version of this standard text. For more information, refer to 'Linking to OMIM over the Internet' later in this section.

Label	Description
Birth Defects Code	Each syndrome is cross referenced to the Birth Defects Encyclopaedia published by the Center for Birth Defects Information Services, Inc., USA.
Proposed Gene	The proposed chromosomal localisation of a related gene (when know).
Other Age Ranges	If a syndrome is described at a number of age ranges, these age ranges are displayed as links. You can click on a link to see the description of the associated age range.
Synonyms	Any alternative terms used for this syndrome.
Patients	Clinic patients that have been diagnosed with this syndrome. You can click on a link to go the description for the associated patient.
Comments	An overview of the syndrome that includes the following: <ul style="list-style-type: none"> • Clinical features • Differential diagnoses; similarities to other syndromes • Radiology • Genetic heterogeneity or clinical sub-types that may occur
References	Text or published articles related to the syndrome.

Label	Description
Traits	<p>The traits that are associated with this syndrome.</p> <p>Note: Syndrome descriptions are drawn from a cross-section of the literature. To maximise the probability of retrieval in a search, all traits reported in the literature are included in a syndrome description. A syndrome is an abstraction—individual patients do not manifest all the features of a syndrome.</p>
Search	<p>List of the traits you have included in your search criteria. The traits that matched are highlighted in bold.</p>

Linking to OMIM Over the Internet

When evaluating a syndrome in POSSUM, you can link (over the Internet) to its reference in the Online Mendelian Inheritance in Man (OMIM). You can establish this link from a syndrome description or from a search results list.



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Click on the MIM number to access the reference.

Note: If you do not have an Internet connection, you will not be able to link to OMIM.

Refer to the OMIM information for the syndrome you are currently working with. You can also follow links and move around through the OMIM web site. Use the Back and Forward buttons to move between pages or snapshot a page if you want to keep it on your desktop.

Looking at Clinic Patients

A patient description includes the following information:

Label	Description
Age	The age at which the photographs were taken.
Date of Birth	The date of birth enables you to calculate the current age of the patient.
Sex	The sex of the patient. This can be male, female or unknown.
Diagnosis Confirmed	Displays 'Yes' or 'No' depending upon whether the diagnosis has been confirmed.
Autopsy Performed	Displays 'Yes' or 'No', depending upon whether an autopsy has been performed.
Comments	Provides the following information about a patient <ul style="list-style-type: none"> • Relevant family history • Results of investigations • Special features that should be noted

Label	Description
Diagnoses	<p>The candidate diagnoses for the patient. 75% of patients in POSSUM have a confirmed diagnosis. The remaining 25% of cases are dysmorphic or have birth defects to a degree that suggests they might have a diagnosable syndrome.</p> <p>By making these cases available to other clinical geneticists, we hope to accelerate the recognition of new syndromes and the diagnosis of patients.</p> <p>Click on a syndrome name to see its description displayed in the POSSUM Browser.</p>
Traits	<p>A list of the traits associated with this patient.</p> <p>Because a clinic patient description is for an individual at a particular stage of development, the list of traits is shorter than those associated with a specific syndrome.</p> <p>The traits associated with a patient represent the features that were apparent at the stage the photographs were taken. The traits may be the same at different ages because the features have changed in degree not type.</p>
Search	<p>The list of traits you used as your search criteria. The traits that matched this patient are highlighted in bold.</p>

Printing Syndrome or Patient Descriptions

You can print a hardcopy of a syndrome or patient description. To do this:

1. Access the syndrome or patient description in the POSSUM Browser.
2. Choose Print from the File menu.
3. Choose the required options on the Windows Print dialog box.
4. Choose OK.

Note: To print POSSUM descriptions, you need an Internet web browser installed on your computer.

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