

# Phenotip tutorial

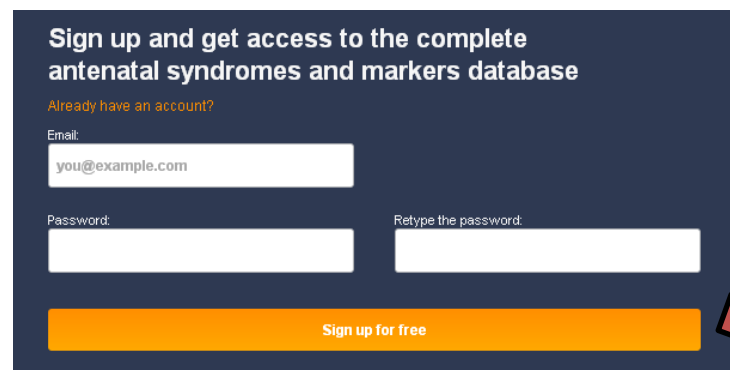
## Introduction

Phenotip is a free, web-based, tool to help diagnosing fetal syndromes antenatally. This website is a searchable database of ultrasound markers (>1000 markers included) and fetal syndromes. Functionality and details have been described in a methodologic [paper](#) and the tool is available at [www.phenotip.com](http://www.phenotip.com). In comparison to other syndrome search tools, Phenotip is only based on fetal findings and not on findings that are only diagnosable postnatally or that appear later in life (such as failure to thrive etc.).

## How to use Phenotip ?

### First time users

Direct your internet browser to [www.phenotip.com](http://www.phenotip.com)

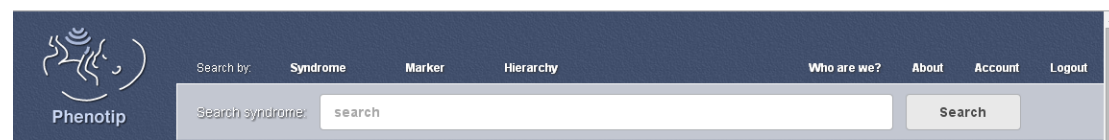


First time users need to sign up. The process is simple – all you need to do is to fill the highlighted boxes and press the “Sign up for free” button. You will get an email confirming your registration.

Next time you get to the welcome page, press the **Login** button on the top right of the screen.

### Returning users

After your successful login, the following search screen will appear:

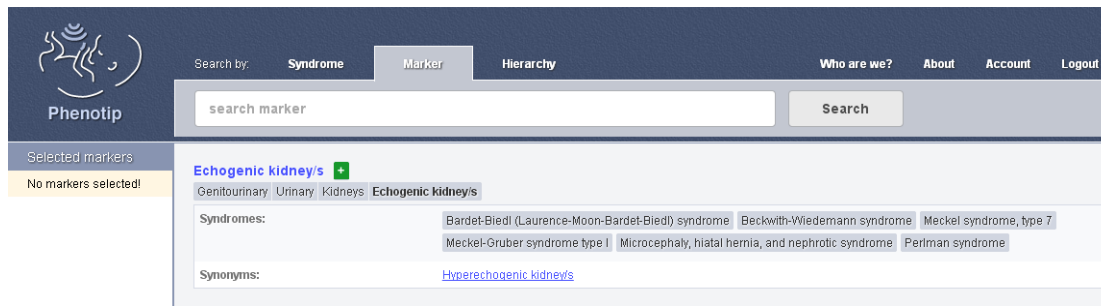


If you want to **find a specific syndrome** by name, press on the **Syndrome** button, and type the syndrome name in the search box.

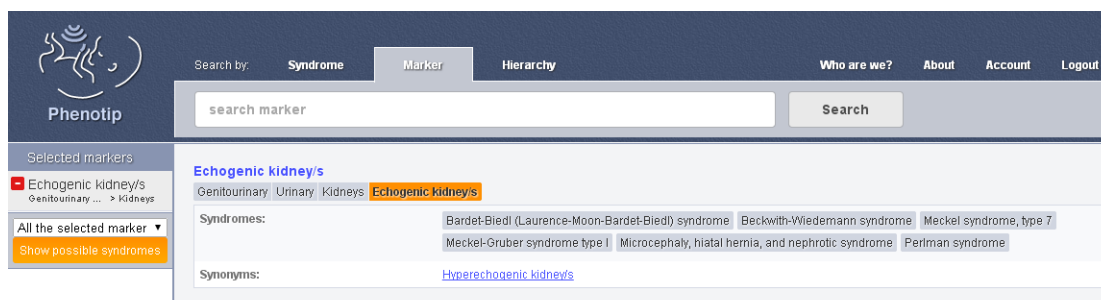
If you want to **find a differential diagnosis** for a given set of sonographic markers, you may choose to search by either **Marker** or by **Hierarchy**.

**Marker**

If you chose **Marker**, start typing a marker's name in the search box. The system will autocomplete after you typed a few letters and recognizes synonyms (eg. Echogenic kidneys and Hyperechogenic kidneys). When you select a marker, it will appear on the screen with the syndromes it is known to be associated with.



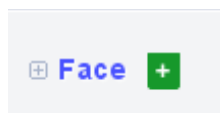
If you want to select that specific marker, hit the **+** sign to the right of the marker's name. It will then appear on the left side of your screen under the “selected markers” heading.



You can repeat the process until you finished adding all the markers you have identified.

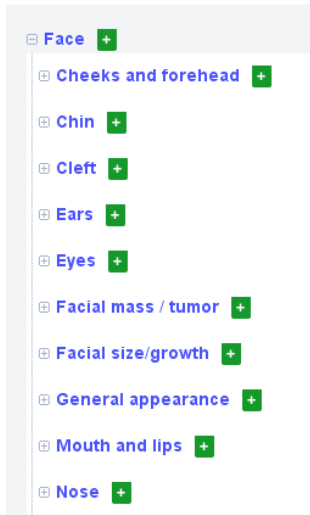
Alternatively, you can choose a marker from an ordered list of markers by clicking the **Hierarchy** tab. **Hierarchy** allow you to choose markers from a branching tree of markers organized by organ, and then by increasing levels of resolution.


For example:




Clicking the **+** sign will select face as a marker, so that all syndromes with any kind of sonographic facial feature will be included in your search.

Clicking on the **+** sign (to the left of the node name) or on the blue node name will open the next level of resolution:

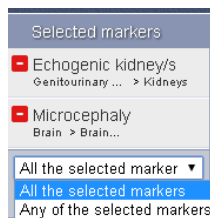


You can continue opening levels until you reach your marker of interest. Click  to add the marker to the 'selected markers' list.

You can remove markers from your list by clicking .

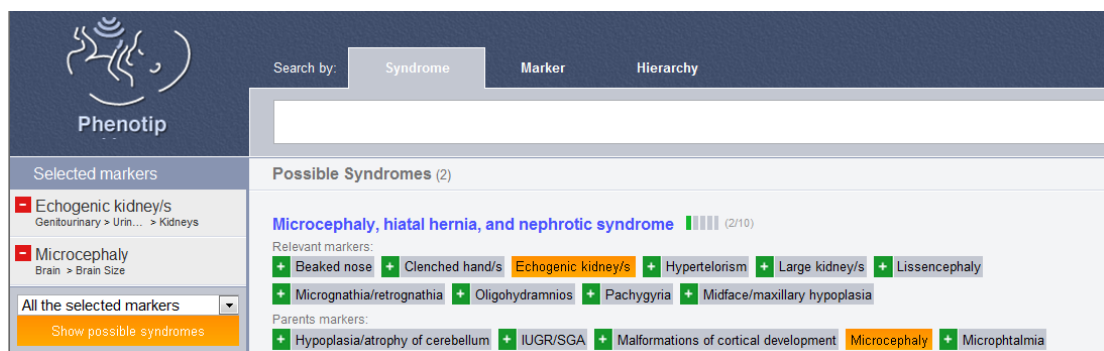
### Ready to get the differential diagnosis of all possible syndromes?

Once you finished adding all markers of interest, choose whether you want find syndromes with "*all selected markers*" or "*any of the selected markers*" from the button at the bottom of the 'selected markers' list on the left hand side of the screen.



The first option will filter the syndrome database for syndromes that include all markers, while the latter option will give all syndromes with one or more markers from the list, ordered by probability of relevance.

Once you hit the 'show possible syndromes' button, a list of candidate syndromes will appear. The sonographic markers you chose appear in orange. For each syndrome however, other described markers will also be visible:



By clicking the syndrome name (in blue), further details will appear, such as links to further information:

Selected markers

+ Echogenic kidney/s  
Genitourinary > Urin... > Kidneys

+ Microcephaly  
Brain > Brain Size

All the selected markers

Show possible syndromes

[Meckel-Gruber syndrome type](#) ||||| (2/36)

Relevant markers:

+ Adrenal agenesis

+ Agnesis of vermis

+ Anal atresia

+ Aortic valve stenosis

+ Arnold-Chiari malformation

+ ASD

+ Bell shaped thoracic cage

+ Clinodactyly

+ Coarctation of aorta

+ Collapsed bladder

+ Dandy Walker malformation

+ Duplex collecting system

+ Echogenic kidney/s

+ Horseshoe kidney

+ Hypertelorism

+ Hypoplastic aorta

+ Hypoplastic bladder

+ Hypoplastic lung/s

+ Hypospadias/tulip sign

+ Hypotelorism

+ Large kidney/s

+ Microcephaly

+ Micrognathia/retrognathia

+ Multicystic dysplastic kidney/s

+ Nuchal edema/fold/increased NT/cystic hygroma

+ Omphalocele

+ Polycystic (enlarged echogenic) kidney/s

+ Postaxial polydactyly of fingers

+ Posterior encephalocele

+ Pulmonary valve stenosis

+ Single umbilical artery

+ Small kidney/s

+ Talipes

+ Thick placenta

+ Undescended testes

+ Oligohydramnios

Parents markers:

+ Agnesis of corpus callosum

+ Ambiguous genitalia

+ Brain

+ Cleft in hard palate

+ Ears

+ Encephalocele

+ Heart

+ Holoprosencephaly

+ Hypoplasia/atrophy of cerebellum

+ IUGR/SGA

+ Microcephaly

+ Microphthalmia

+ Polydactyly of hand/s

+ Renal agenesis

+ Short limbs

+ Syndactyly of fingers

+ Ventriculomegaly

+ VSD

Incidence: 1/142860 — -

Involved genes:  
(gene — definition — chromosome)
MKS1 — 17q22, TMEM216 — 11q12.2

Inheritance mode: AR

Other online databases: [OrphaNet](#), [OMIM](#), [Geneva Foundation](#)

Links: [www.sonoworld.com](http://www.sonoworld.com)

Good luck !!

The Phenotip team