

# Tutorial for testing Dr. Rachael

## Introduction

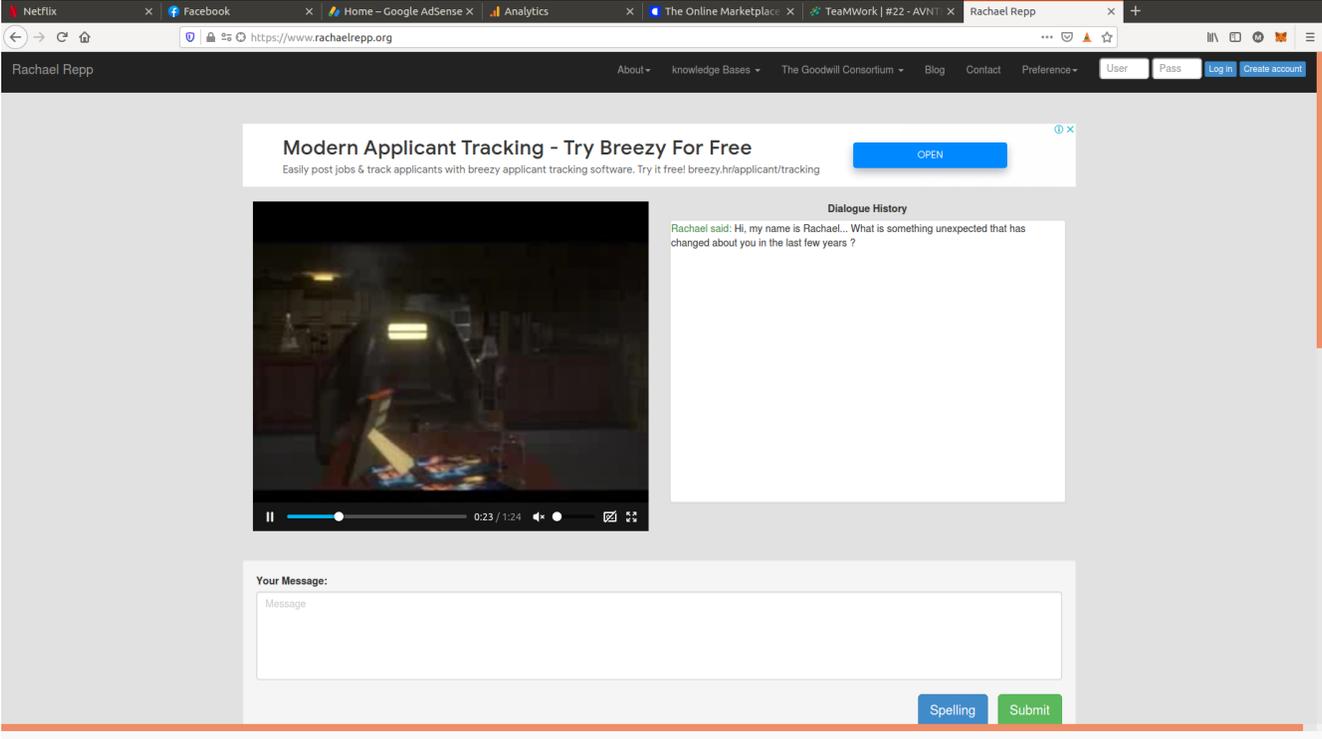
Dr. Rachael is able to analyze symptoms associated with 600 rare diseases currently. Please, look at the “rare diseases” tab for a full list. When asked to make suggestions, it takes all the conditions with higher than a 5% probability (though we will take this threshold up to 10% in the future) and displays them to the user. The user must click on the correct condition if known, or tick “none of the above” and enter the correct diagnosis if known, prior to pressing “Save”.

According to a recent study by NORD, more than 40% of rare disease patients have been misdiagnosed at least once on the way to a successful diagnosis so validating the diagnosis by investigating possible other explanations for the identified symptoms is something that is useful and will also help us validate Dr. Rachael’s performance in the first instance.

## Step 1

First of all, go to our development site: [www.rachaelrepp.org](https://www.rachaelrepp.org)

You can chat with the general version of Rachael for a while and see what she is like. Her knowledge base is approximately 100,000 words long.



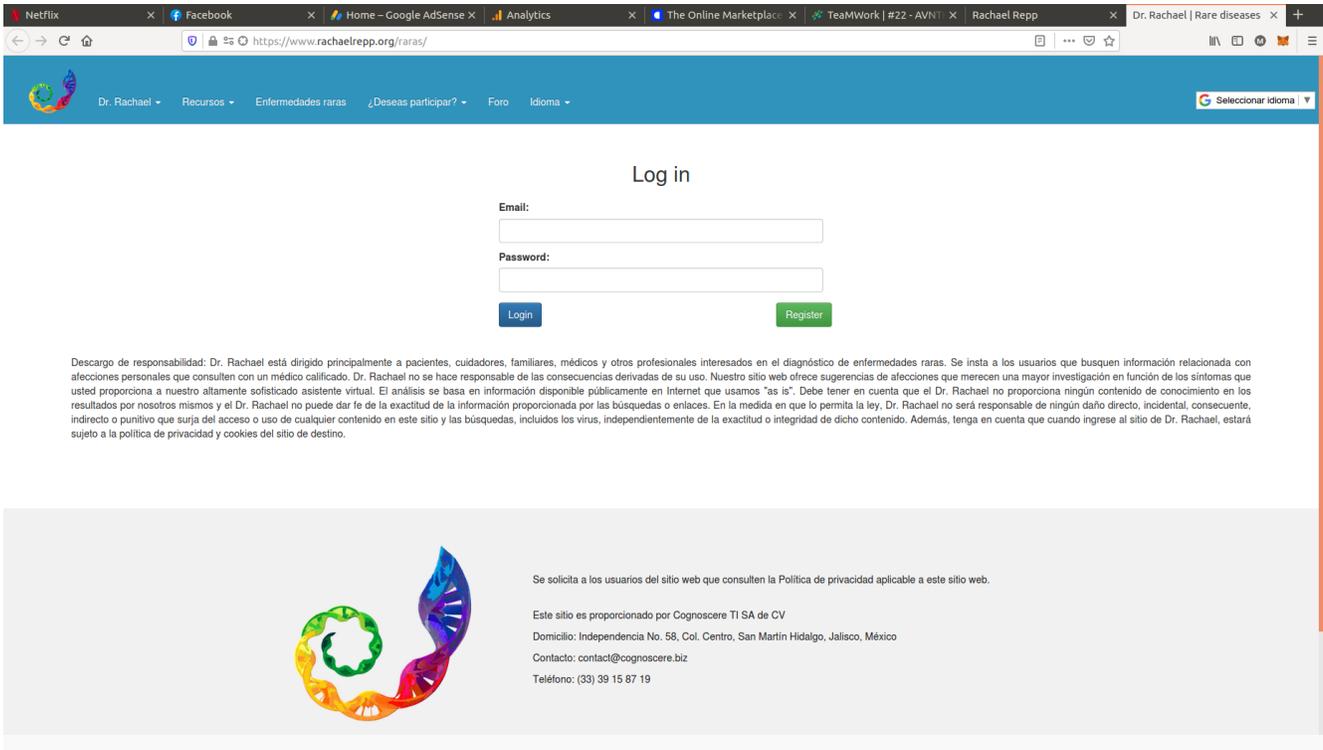
The screenshot shows a web browser window with the URL <https://www.rachaelrepp.org>. The page features a navigation bar with links for "About", "knowledge Bases", "The Goodwill Consortium", "Blog", "Contact", and "Preference". There are also buttons for "User", "Pass", "Log in", and "Create account".

The main content area includes a promotional banner for "Modern Applicant Tracking - Try Breezy For Free" with an "OPEN" button. Below this is a video player showing a person in a dark setting. To the right of the video is a "Dialogue History" section with the text: "Rachael said: Hi, my name is Rachael... What is something unexpected that has changed about you in the last few years ?".

At the bottom, there is a "Your Message:" section with a text input field labeled "Message" and "Spelling" and "Submit" buttons.

## Step 2

Select “Knowledge bases” → “Rare Diseases” A login page will appear



The screenshot shows a web browser window with multiple tabs open. The active tab is 'Dr. Rachael | Rare diseases'. The address bar shows the URL 'https://www.rachaelrepp.org/raras/'. The website header is blue and contains a logo on the left, navigation links for 'Recursos', 'Enfermedades raras', '¿Deseas participar?', 'Foro', and 'Idioma', and a language selection dropdown on the right. The main content area is white and features a 'Log in' heading. Below the heading are two input fields: 'Email:' and 'Password:'. There are two buttons: a blue 'Login' button and a green 'Register' button. Below the login form is a small paragraph of disclaimer text in Spanish. At the bottom of the page, there is a footer with a colorful DNA helix logo on the left and contact information on the right, including the website name 'Cognoscere TI SA de CV', address, email, and phone number.

Dr. Rachael | Recursos | Enfermedades raras | ¿Deseas participar? | Foro | Idioma | Selecionar idioma

### Log in

Email:

Password:

[Login](#) [Register](#)

Descargo de responsabilidad: Dr. Rachael está dirigido principalmente a pacientes, cuidadores, familiares, médicos y otros profesionales interesados en el diagnóstico de enfermedades raras. Se insta a los usuarios que busquen información relacionada con afecciones personales que consulten con un médico calificado. Dr. Rachael no se hace responsable de las consecuencias derivadas de su uso. Nuestro sitio web ofrece sugerencias de afecciones que merecen una mayor investigación en función de los síntomas que usted proporciona a nuestro altamente sofisticado asistente virtual. El análisis se basa en información disponible públicamente en Internet que usamos "as is". Debe tener en cuenta que el Dr. Rachael no proporciona ningún contenido de conocimiento en los resultados por nosotros mismos y el Dr. Rachael no puede dar fe de la exactitud de la información proporcionada por las búsquedas o enlaces. En la medida en que lo permita la ley, Dr. Rachael no será responsable de ningún daño directo, incidental, consecuente, indirecto o punitivo que surja del acceso o uso de cualquier contenido en este sitio y las búsquedas, incluidos los virus, independientemente de la exactitud o integridad de dicho contenido. Además, tenga en cuenta que cuando ingrese al sitio de Dr. Rachael, estará sujeto a la política de privacidad y cookies del sitio de destino.

Se solicita a los usuarios del sitio web que consulten la Política de privacidad aplicable a este sitio web.

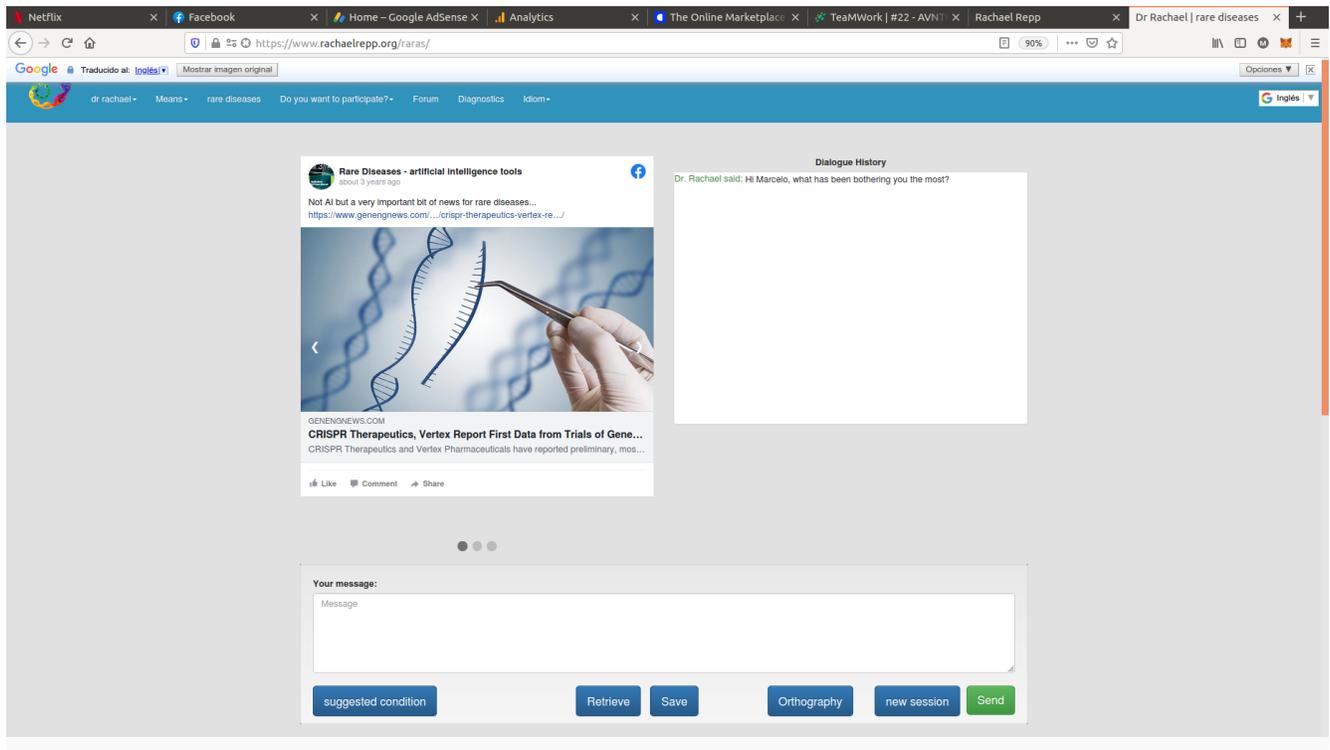
Este sitio es proporcionado por Cognoscere TI SA de CV  
Domicilio: Independencia No. 58, Col. Centro, San Martín Hidalgo, Jalisco, México  
Contacto: [contact@cognoscere.biz](mailto:contact@cognoscere.biz)  
Teléfono: (33) 39 15 87 19

If this is your first time, you will need to register first. This is free and only done to assign a name to your contributions and have a contact e-mail

The system currently runs in English, Spanish and French only. Please, select the correct language for you.

## Step 3

Once logged in, the main page will appear.



The memory knowledge base has three components: a general knowledge and personality knowledge base, a general medicine knowledge base (which needs a fair amount of additions) and a knowledge base of the most common symptoms associated to 600 of the rare diseases with the higher incidence, though there are some special cases such as diseases that now have treatment and did not have a cure in the past, and some diseases that were originally included for early testing purposes.

## Step 4

We mostly developed the knowledge base using information from NORD and GARD so for testing purposes, we suggest you avoid these sources but look for your own, the patients symptoms or obtain some symptoms from other bibliographical sources. The “rare diseases” tab has a list of the rare diseases we have considered so far. These are of three type: on the left hand side are the diseases that Dr. Rachael currently is able to look out for, the middle column refers tot hose being considered currently (empty) and the right hand side column includes the conditions that we have decided not to include for a variety of reason; mostly that there are a number of rare diseases that are rare but actually not hard to identify. Note you need to enter spelling and punctuation correctly.

Just as an example, Barth syndrome was chosen and typical symptoms are listed below, each was entered sentence by sentence to the virtual assistant.

- The child has heart problems
- The child has arrhythmia.
- The child suffers from shortness of breath.
- The child has mouth ulcers.
- The child has frequent infections.
- The child has weak muscles.
- There is curvature of the spine.
- The child has a round face with prominent chin and full cheeks.
- The child has some learning disability.
- The child has large ears and deep set eyes.

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Rachael Repp

Dr Rachael | rare diseases

Google

Traducido al: **inglés**

Mostrar imagen original

Opciones

dr rachael

Means

rare diseases

Do you want to participate?

Forum

Diagnostics

Idiom

inglés

**Rare Diseases - artificial intelligence tools**

About 3 years ago

Not AI but a very important bit of news for rare diseases...

[https://www.genengnews.com/.../crispr-therapeutics-vertex-re...](https://www.genengnews.com/.../crispr-therapeutics-vertex-re.../)

GENENGENEWS.COM

**CRISPR Therapeutics, Vertex Report First Data from Trials of Gene...**

CRISPR Therapeutics and Vertex Pharmaceuticals have reported preliminary, mos...

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Meld Diff Viewer

**Dialogue History**

Dr. Rachael said: Hi Marcelo, what has been bothering you the most?

Marcelo said: There is curvature of the spine.

Dr. Rachael said: Scoliosis is an abnormal curvature of the spine that causes the body to roll sideways.

Marcelo said: The child has mouth ulcers.

Dr. Rachael said: Tell me more about your symptoms.

Marcelo said: The child has arrhythmia.

Dr. Rachael said: What other symptoms have you had?

Marcelo said: The child has weak muscles.

Dr. Rachael said: Tell me more about your symptoms.

Marcelo said: The child has large ears and deep set eyes.

Dr. Rachael said: What other symptoms have you had?

Marcelo said: The child has some learning disability.

Dr. Rachael said: Tell me more about your symptoms.

Marcelo said: The child has a round face with prominent chin and full cheeks.

Dr. Rachael said: Tell me more about your symptoms.

Your message:

Message

suggested condition

Retrieve

Save

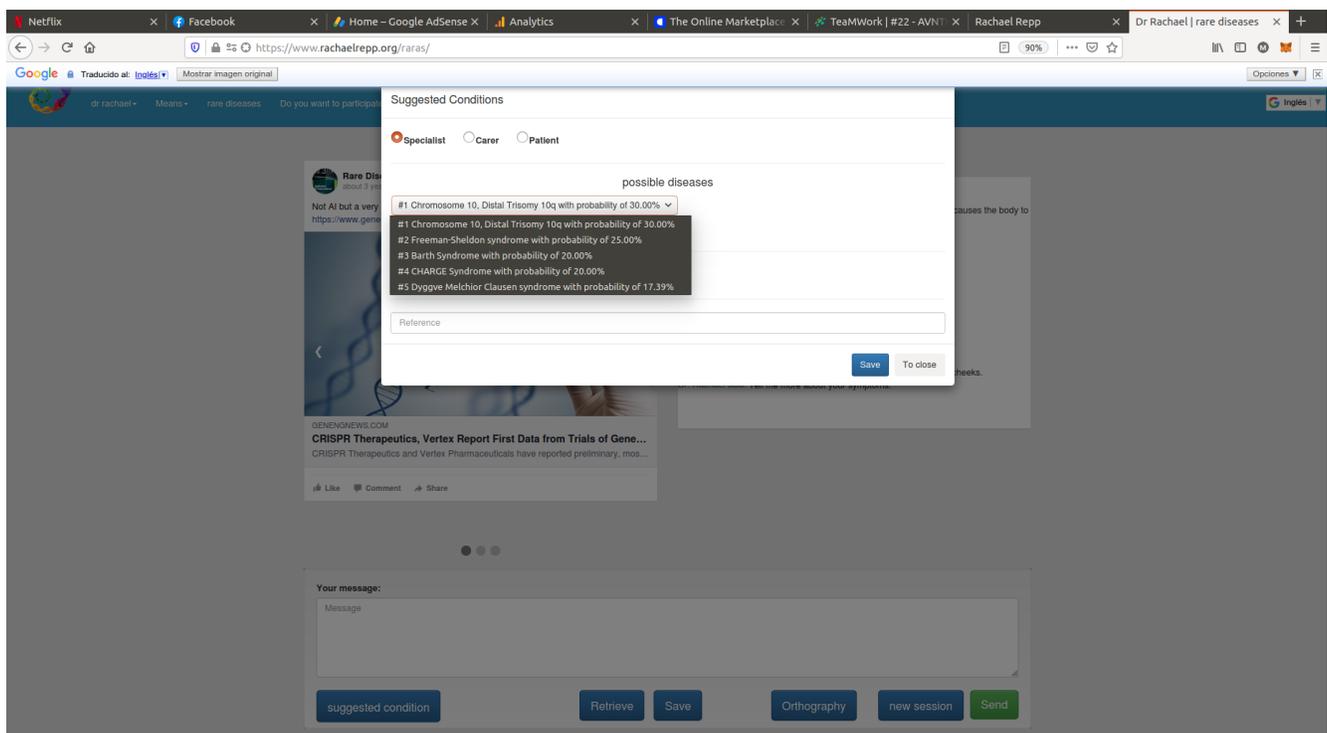
Orthography

new session

Send

- A few points are worthy of mention here: The first comment is fine, a general medical comment... but all the others are given because Dr. Rachael does not have enough comments that are relevant... any relevant general comment suggestions are welcome!
- The order and amount of symptoms matter, the more details you can put in the better... hence it is important to be able to save a session and be able to come back to it and keep adding symptoms.
- Sometimes, the general knowledge module responds and the Dr. Rachael says something totally irrelevant... it does not affect the results but just the interaction but clearly we need to work on that.

At any time the user can press “suggested condition” and a results window will appear showing the possible diseases being considered; currently including all those above a 10% probability.



Working out the probability is actually not straight forward. Currently, we are taking the probability to be the number of matching symptoms divided by the average number of symptoms in our disease list (21). There are some diseases that only have 6 symptoms... others have 25... so if we divided the number by the total number of symptoms and say you had 3 symptoms.. you'd have a probability of 50% in the first case and 15% in the second... we found that dividing by the average gave us more robust consistent results... we could multiply by the incidence of two diseases but... those statistics are not very reliable and change a lot between countries... but may be we could just try in the near future; we are currently compiling this list.

Another substantial problem is that there are families of only a few diseases that can be identified by having just 3-4 certain symptoms... but there are some families of symptoms that are associated with may be 200 diseases... so we are thinking of changing this calculation so that it shows all the diseases that have more than a 10% probability by groups, but currently we are using 10%, which actually makes that list very long at times. These are all the conditions the system considers could be potentially associated with the symptoms provided.

**If the correct disease is on the list, the user must click on it and it will be taken to the top of the list which is the one saved; by default the disease with the highest probability is placed at the top.**

If the system fails and the correct rare diseases is not on the list, then the user needs to tick the “none of the above” box and enter the correct diagnosis in the space provided.

Failure can be due to a variety of reasons. Sometimes the sentences in Dr. Rachael’s knowledge base need to be broken down into simpler sentences. Sometimes there may be some co-morbidity that means the user entered symptoms that have nothing to do with the main condition but are caused by it or simply happen to apply to the person for other reasons, etc. We are currently reviewing the rare diseases knowledge base and are about half way though.

In order to get some indication of effectiveness of the system under different conditions, we distinguish between carer, patient or specialist. So, before the “save” button is pressed, please click on the appropriate buttons. Each sort of user might employ a different vocabulary and it may be Dr. Rachael is sensitive to that; though she has a 35,000-words scientific dictionary available.

Also, it would be useful to know, in the cases where a patient’s actual symptoms are being described, if the diagnosis being taken as correct was provided by a genetic test or after a clinical analysis.

A “reference” text space is provided for the user to enter a private code or ID number to enable the user to identify the patient associated with the information later on, and privacy is respected.

Finally, the “save” button will add the result to our test database to which we will apply some data science technique to try and help us identify errors, areas of improvement and the rate of successful identification currently being obtained. Just as an indication, if only relevant symptoms are included, the systems yielded a success rate of 75% and if at least half of the symptoms corresponded to the underlying condition then a success rate of 50% has been obtained during the initial tests, which are based on only 300 rare diseases. The results of each test are added and will appear in the database window which you can see by pressing “diagnostics” in the upper menu:

DATE	USERNAME	STATUS	REFERENCE	RARE DISEASES	DIAGNOSTICS	TYPE	EXPECTED DIAGNOSIS
2020-10-02	ladislaus	specialist	IMSS12334	<ul style="list-style-type: none"> <li>Fragile X Syndrome with probability of 15.33%</li> <li>Rett syndrome with probability of 9.33%</li> <li>Pulmonary Arterial Hypertension with probability of 3.33%</li> </ul>	Fragile X Syndrome	Clinical	Right
2020-10-05	ladislaus	Career	NF00123	<ul style="list-style-type: none"> <li>Fragile X Syndrome with probability of 15.33%</li> <li>Rett syndrome with probability of 9.33%</li> <li>Pulmonary Arterial Hypertension with probability of 3.33%</li> </ul>	Rett syndrome	Clinical	Right
2020-10-05	ladislaus	Patient	IMSS0001	<ul style="list-style-type: none"> <li>Fragile X Syndrome with probability of 15.33%</li> <li>Rett syndrome with probability of 9.33%</li> <li>Pulmonary Arterial Hypertension with probability of 3.33%</li> </ul>		Genetic	I do not identify
2020-10-05	Marcelo	Career	IMSS001	<ul style="list-style-type: none"> <li>Fragile X Syndrome with probability of 15.33%</li> <li>Rett syndrome with probability of 9.33%</li> <li>Pulmonary Arterial Hypertension with probability of 3.33%</li> </ul>	Rett syndrome	Clinical	Right
2020-10-25	Marcelo	Career		<ul style="list-style-type: none"> <li>ADNP Syndrome with probability of 103.33%</li> <li>Leber's Congenital Amaurosis with probability of 18.67%</li> <li>Joubert Syndrome with probability of 16.00%</li> </ul>	Joubert Syndrome	Clinical	Right
2020-11-08	ladislaus	Career	6 symptoms mentioned.	<ul style="list-style-type: none"> <li>Klippel-Trenaunay Syndrome with probability of 30.00%</li> <li>Essential Thrombocythemia with probability of 15.00%</li> <li>Oculo-Auriculo-Vertebral Spectrum with probability of 15.00%</li> </ul>	Klippel-Trenaunay Syndrome	Clinical	Right
2021-03-14	ladislaus	Career		<ul style="list-style-type: none"> <li>Freeman-Sheldon syndrome with probability of 50.00%</li> <li>Chromosome 10, Distal Trisomy 10q with probability of 15.00%</li> <li>Spondyloepiphyseal Dysplasia, Congenital with probability of 8.70%</li> </ul>	Freeman-Sheldon syndrome	Clinical	Right

The current conditions listing is shown below:

Search...

Conditions contemplated by Dr. Rachael		
Currently included (600)	Under consideration (550)	Not included (52)
Crohn's Disease		Great vessels transposition
Acromegaly		Proteus syndrome
Lyme disease		neurofibromatosis
Meningitis		Monoclonal Gammopathy of Undetermined Significance
Porphyria		Oculocutaneous albinism
Dercum's Disease		streptococci
multiple sclerosis		Meyer Dysplasia
Neuromyelitis Optica (Devic Disease)		Staphylococci
Pulmonary Arterial Hypertension		Fitzsimmons Watson Mellor syndrome
hemophilia		fibrochondrogenesis
Guillain-Barre syndrome		pneumococci
Madelung's disease		astroblastoma
Marfan syndrome		Actinic lichen planus
Parsonage Turner Syndrome		aldred syndrome

[Add Row](#) [Delete Row](#)

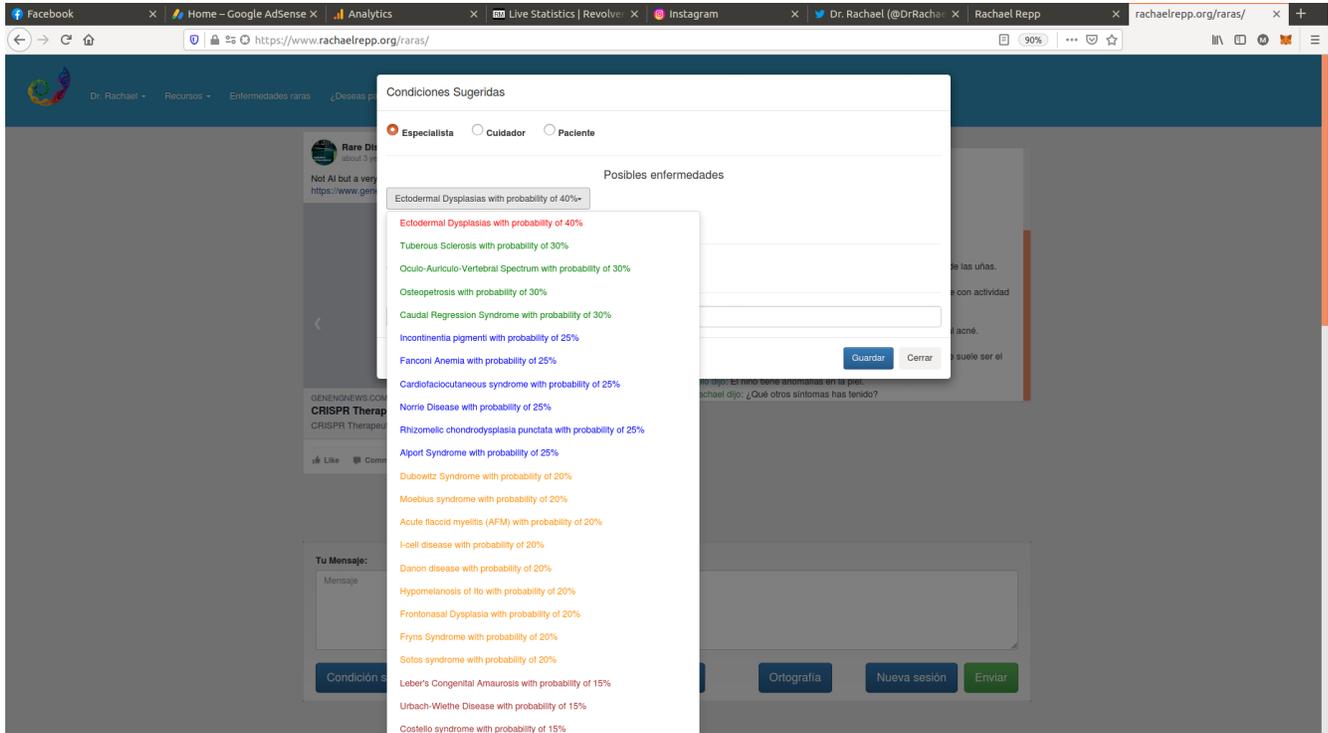
## Step 5

If the user wishes to test a number of cases, the memory can be wiped clean and a new case investigated by pressing the “new session” button. A second test run was done, this time in Spanish in a case of tuberous sclerosis:

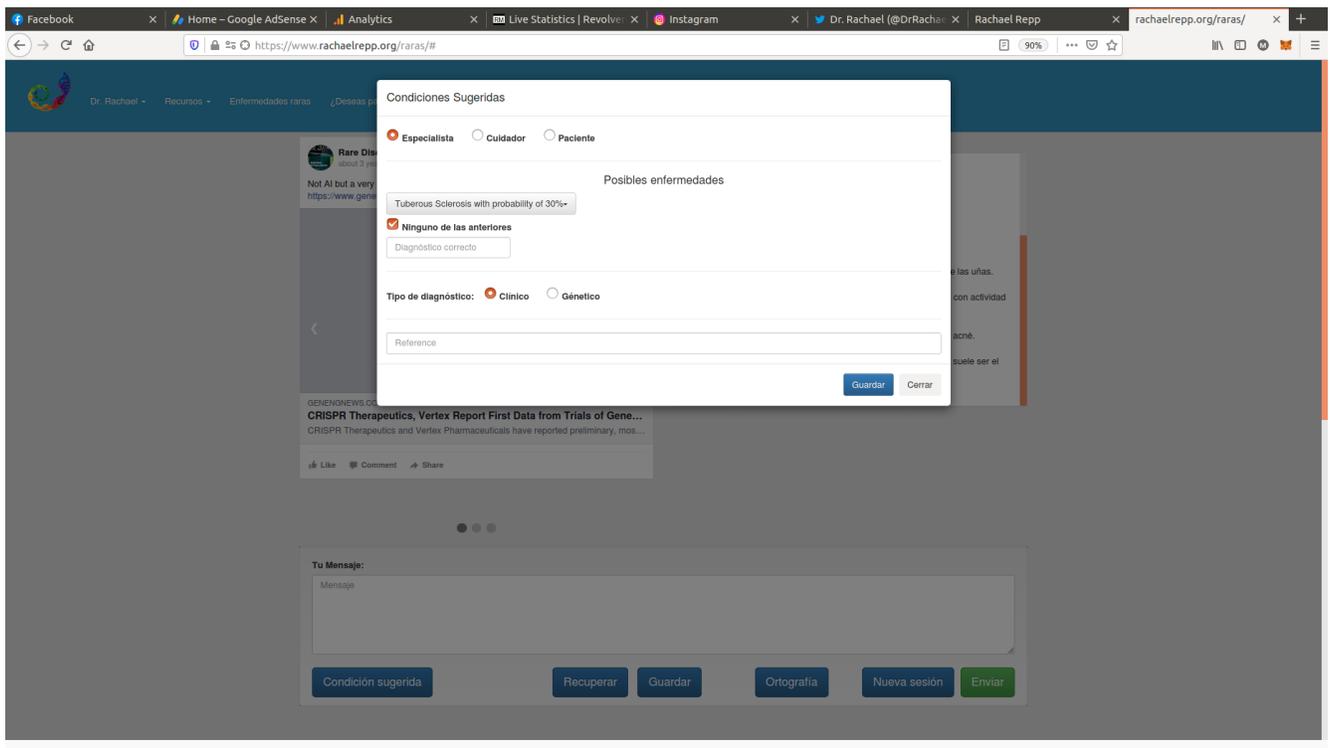
- El niño tiene trastorno del espectro autista.
- El niño tiene crecimientos que aparecen como manchas blancas en la retina.
- El niño tiene problemas de comportamiento.
- El niño tiene trastornos del sueño.
- El niño tiene dolores de cabeza a menudo.
- El niño tiene problemas de aprendizaje.
- El niño tiene manchas de piel de color claro.
- El niño tiene trastorno de hiperactividad.
- El niño tiene protuberancias rojizas debajo y alrededor de las uñas.
- El niño tiene tos y dificultad para respirar, especialmente con actividad física o ejercicio.
- Han aparecido crecimientos faciales que se asemejan al acné.
- El niño tiene anomalías en la piel.

The screenshot displays a web browser window with multiple tabs. The active tab is 'rachelrepp.org/raras/'. The page content shows a chat interface for 'Rare Diseases - artificial intelligence tools'. The chat history on the right side of the screen shows a conversation in Spanish. The user 'Marcelo' provides a list of symptoms, and 'Dr. Rachael' responds with diagnostic suggestions. At the bottom of the chat window, there is a text input field labeled 'Tu Mensaje:' and several buttons: 'Condición sugerida', 'Recuperar', 'Guardar', 'Ortografía', 'Nueva sesión', and 'Enviar'.

Note that when you get “undefined” either there is a problem with processing the sentence, the language detection or the system is down and you have to send me a message (Whatsapp: +523339554580, skype: gallanzi) to pull it back up. Try a few sentences and if you keep getting undefined it is because the system is down.



In this case, the correct diagnosis appears in second place with a probability of 30% against the system's favored diagnosis of one of the ectodermal dysplasias with 40%. So The user must click on the correct diagnosis and this condition will be moved to the top position and this test run can then be saved:



In order to test the system well, ideally users should choose a diseases randomly from the list and try getting the symptoms from other Internet pages and entering these symptoms in using different words and grammar.

Later on, we intend the user to also fill in a electronic medical record and the results of this initial step will be entered first before the user describes the symptoms. Here is a draft of the page for entering the electronic medical record:

The screenshot shows a web browser window with the URL <https://www.rachaelrepp.org/raras/general-info>. The page title is "General information" and the breadcrumb is "Start > General information of the interested party". The form is titled "Information of the patiente" and includes the following fields:

- \* Names
- \* Surnames
- \* Email
- Address
- Sex (dropdown menu)
- Instagram
- \* WhatsApp
- Telegram
- \* Home Phone
- mm / dd / yyyy

A "Finish Registration" button is located at the top right of the form area. A "to register" button is located at the bottom left of the form area. The browser's address bar shows the URL and the page is zoomed in at 90%.

We still need to finish linking to the associated MySQL database, ask the user to fill in the details and may be choose relevant things and transfer them to Dr. Rachael up front before the person adds further symptoms.