RareConnect.ORG
A PARTNERSHIP OF EURORDIS AND NORD

Supporting European Federations

Rob Pleticha, Online Communities Manager, EURORDIS

RareConnect in numbers I

- **32** communities live
- **5** communities in development for 2012: Porphyria, Propionic Acidemia, Univentricular heart, Ring Chromosome 11, Charcot Marie Tooth
- **3,000** registered members

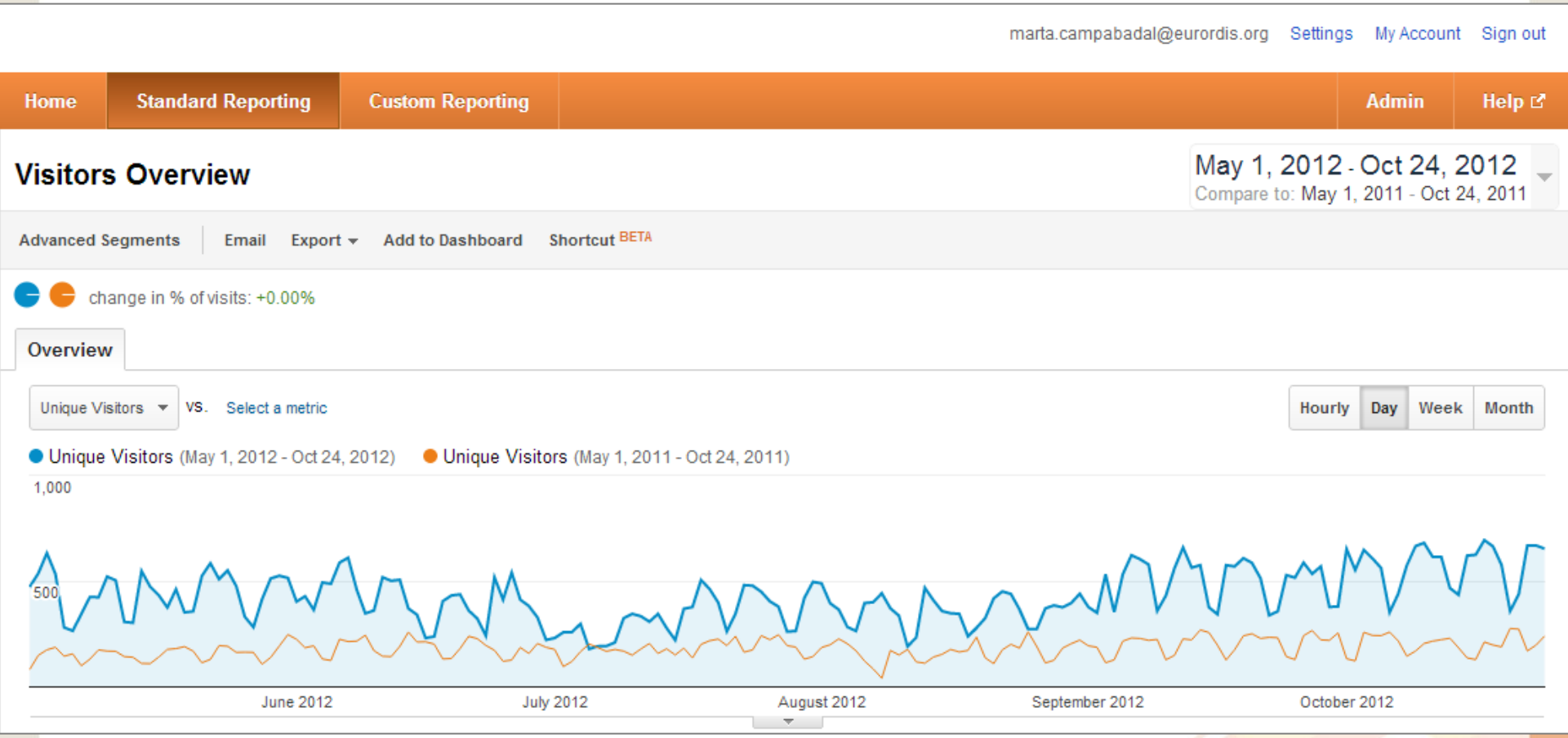
RareConnect in numbers II

- **93** volunteer moderators
- **239** international patient group partners
- **40,000** words translated monthly on average

Alkaptonuria (AKU)
Alstrom Syndrome
Alternating Hemiplegia
Atypical Hemolytic Uremic Syndrome
Behçet's Syndrome
CAPS
CDG
Coats Disease
Cystinosis
Dravet Syndrome
DysNet
Ehlers-Danlos Syndrome
Epidermolysis Bullosa
Evans Syndrome
Familial Mediterranean Fever
Fibromuscular Dysplasia

Glut1 DS
Hereditary Spastic Paraplegia
Lipoprotein Lipase Deficiency
Mastocytosis And Mast Cell Activation Disorders
Moebius Syndrome
Multiple Myeloma
Multiple System Atrophy
Narcolepsy
Neuroacanthocytosis
Paraneoplastic Neurological Syndrome
Pulmonary Hypertension
Rett Syndrome
Trimethylaminuria
Von Hippel-Lindau
Waldenstrom Macroglobulinemia
WHIM Syndrome (WHIM)

Last 6 months



Oct 24 2011: 240 unique visitors/day
Oct 24 2012: 653 unique visitors/day

150% increase

Last 6 months

	Country / Territory	Visits	% Visits
1.	United States	19,116	22.39%
2.	Italy	10,996	12.88%
3.	France	10,665	12.49%
4.	Germany	7,215	8.45%
5.	Spain	5,600	6.56%
6.	United Kingdom	5,577	6.53%
7.	Canada	2,429	2.84%
8.	Switzerland	1,429	1.67%
9.	Mexico	1,383	1.62%
10.	Argentina	1,306	1.53%

Social media outreach

- To amplify, promote and share your work and group's message (ex. Awareness days)

- To attract new stakeholders, bloggers, isolated patients and families

A few examples….


Understand
Meet
Ciao!
Hola!
Bonjour!
Learn
Connecting Rare Disease Patients Globally
RareConnect
672 likes · 222 talking about this
Non-Governmental Organization (NGO)
Connecting Rare Disease Patients Globally www.rareconnect.org
www.twitter.com/rareconnect
672
About
Photos
Likes
Twitter

RareConnect shared a link.
20 hours ago

Today October 25th is the first day of the International Epidermolysis Bullosa Awareness Week. (October 25-31)

To understand how to live with EB, to learn more about this disease and to meet others visit the Epidermolysis Bullosa community on RareConnect.

Epidermolysis Bullosa, the Community - RareConnect
www.rareconnect.org

Epidermolysis Bullosa (EB) is a group of rare, inherited disorders that affects skin and mucous membranes. The clinical picture

Like · Comment · Share 1

Sue Sheehy, Elina Miaouli and Roxy Foley like this.

Write a comment...

131 people saw this post Promote

RareConnect shared a link.
19 October

Jeff from AHC Kids Foundation, Mirta from American Behcet's Disease Association and Christy from the Cystinosis Research Network will all participate in a panel at the US conference on Rare Diseases happening in Washington D.C.

What do they all have in common? There are communities on RareConnect for these diseases. Visit www.rareconnect.org to find out what other diseases are covered.

Patient Advocates Tell of Struggles in Getting Drugs Developed for Rare Diseases Rare Disease Associ
www.cnbc.com

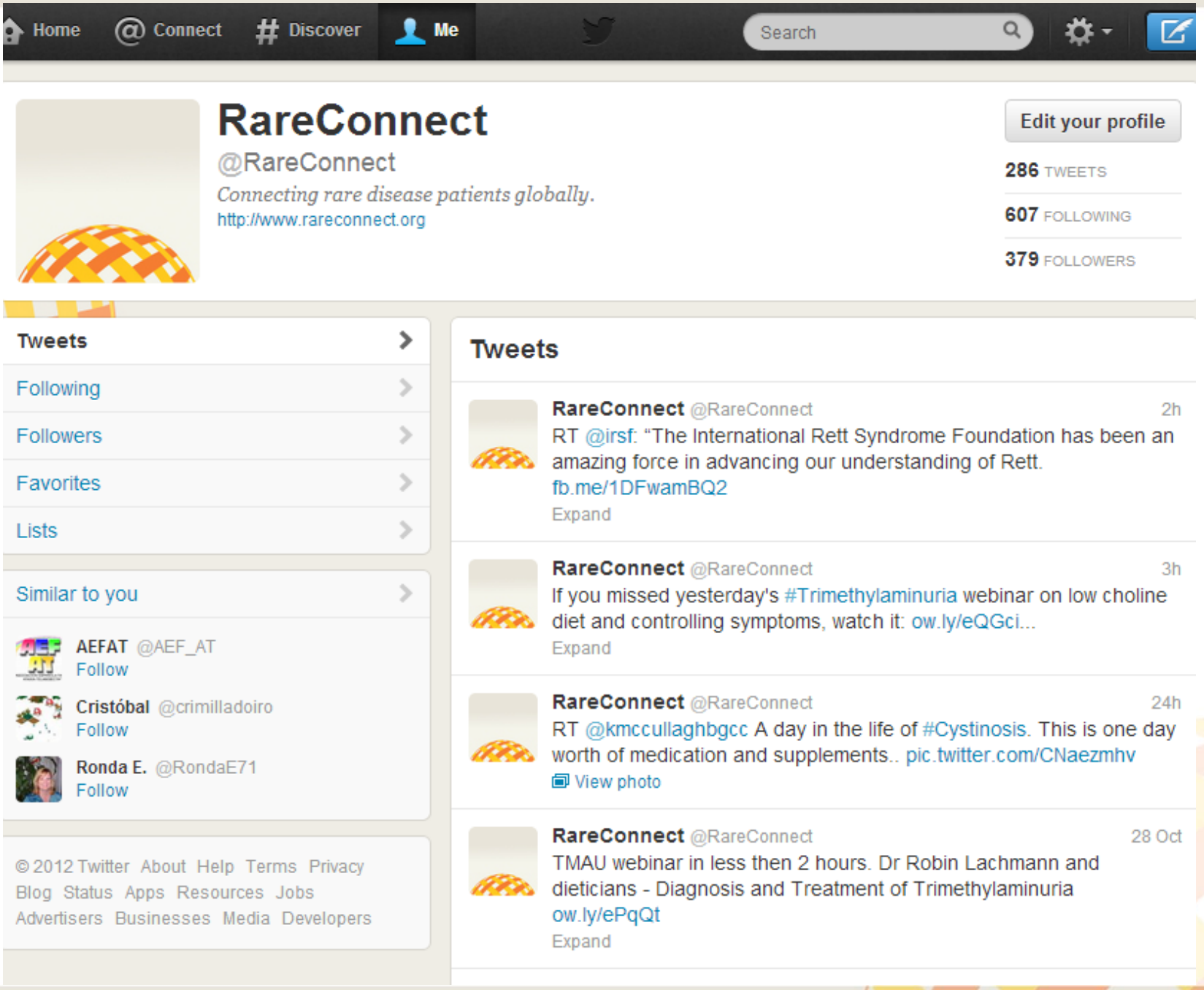
Patient Advocates Tell of Struggles in Getting Drugs Developed for Rare Diseases Rare Disease Associations Perform Own Research, Patient Recruitment, Clinical Trials

Unlike · Comment · Share 13

RareConnect, Diane Kowalik, Mumin Sadiku, Heather Strange and 3 others like this.

Write a comment...

173 people saw this post Promote


Home
Connect
Discover
Me
Search
RareConnect
@RareConnect
Connecting rare disease patients globally.
http://www.rareconnect.org
Edit your profile
286 TWEETS
607 FOLLOWING
379 FOLLOWERS
Tweets
Following
Followers
Favorites
Lists
Similar to you
AEFAT @AEF_AT
Follow
Cristóbal @crimilladoiro
Follow
Ronda E. @RondaE71
Follow
© 2012 Twitter About Help Terms Privacy
Blog Status Apps Resources Jobs
Advertisers Businesses Media Developers
Tweets
RareConnect @RareConnect 2h
RT @irsf: "The International Rett Syndrome Foundation has been an amazing force in advancing our understanding of Rett.
fb.me/1DFwamBQ2
Expand
RareConnect @RareConnect 3h
If you missed yesterday's #Trimethylaminuria webinar on low choline diet and controlling symptoms, watch it: ow.ly/eQGci...
Expand
RareConnect @RareConnect 24h
RT @kmccullaghbgcc A day in the life of #Cystinosis. This is one day worth of medication and supplements.. pic.twitter.com/CNaezmhv
View photo
RareConnect @RareConnect 28 Oct
TMAU webinar in less then 2 hours. Dr Robin Lachmann and dieticians - Diagnosis and Treatment of Trimethylaminuria
ow.ly/ePqQt
Expand

RareConnect @RareConnect 24 Oct

Interested in autoinflammatory disorders? @NOMIDAlliance has an advocacy and updates newsletter you should subscribe to bit.ly/SrkzQw

EDS UK @ehlersdanlosuk 16 Oct

Research questionnaire-Are you over 18? Have you been diagnosed with hypermobility syndrome or EDS Hypermobility? sdu-surveys.herts.ac.uk/hypermobility_…

Retweeted by RareConnect

Bringing patient groups together

- Not only a place for patients and caregivers to exchange

- Shared project is catalyst for starting and maintaining a vibrant European federation

Hereditary Spastic Paraplegia

Dysmelia

Waldenstroms Macroglobulenemia

3rd HSP Congress (10/19-20/2012 Madrid)

by **Tarinda** published about 1 month ago

The Spanish Association of Hereditary Spastic Paraplegia (AEPEF),will hold the 3rd Hereditary Spastic Paraplegia (HSP) Conference "The current state of the study and treatment of Spasticity and associated pathologies" the 19th and 20th of October to celebrate the 10th anniversary of the association's founding. The conference will include the participation of clinical specialists ...

Read more

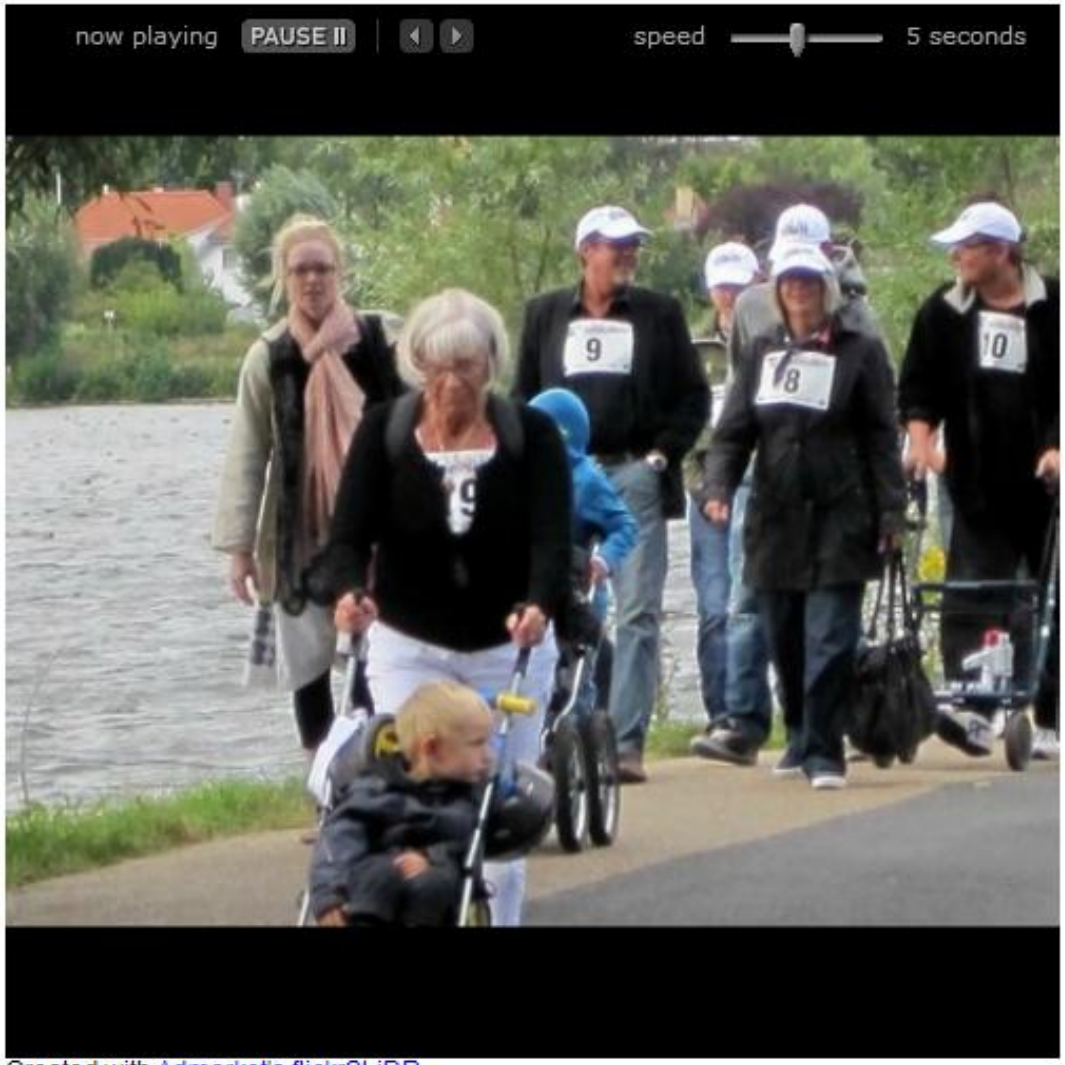
Germany: The Tom Wahlig Foundation for HSP Research

by Tom Wahlig Stiftung published 3 months ago

Video in German language.
View the entire EURORDIS newsletter article on HSP and the Tom Wahlig Foundation in Germany (available in 6 languages).
Dr. Tom Wahlig set ...

"Walk ´n Roll" – September 2012

by Dorthe, HSP Denmark published about 1 month ago



Created with Admarket's flickrSLiDR.

"Foreningen for Ataksi / HSP" arranged a "Walk ´n Roll" event around the Damhus lake on Saturday September 8.

2012 Spastic Paraplegia Foundation Annual Conference

by **franc1s** published about 1 month ago

The theme of the June 2012 Annual Conference of the Spastic Paraplegia Foundation was appropriately called "Monumental Hopes and Dreams for a Cure".

http://www.sp-foundation.org/content/event... Over 120 people attended from 26 states of the United States as well as people from Australia, India, New Zealand and UK. It was held in McLean, Va. in the ...

Read more

Get started

- Send your **interest** to Rob: robert.pleticha@eurordis.org
- Organise **conference call** to involve other groups
- Fill out a **Seeding Document**, gather information from your website
- We create **Test Site**
- **Launch**
- **Promote**

Make it active

- **Join**: http://www.rareconnect.org/en/register
- **Share the link** to the Community on your website, social media, newsletter
- **Encourage your members to join** the international conversation enabled by human translation
- Post **updates** on your group's activities, share your story
- Place **badge** on your website/blog or add RSS feed to your website

Keep it active

- Write a **scientific article summary** and open up the full text via patientINFORM
- Organise a **webinar** with a specialist or researcher
- Create a short **video** interview
- Share **photos** from your recent event
- Invite the RareConnect team to your next **conference**

patientINFORM

ACCESS + INTERPRETATION = UNDERSTANDING + EMPOWERMENT

Access to free full text articles in selected journals

Process:

1. Check with Rob that article is available, request full text version
2. Write summary
3. We create summary page on PH Community with link to abstract
4. patientINFORM works with journals to open up access to full text

De novo mutations in ATP1A3 cause alternating hemiplegia of childhood

Written by **Marcus**, published about 1 month ago.

Summary of article: Erin L Heinzen et al. De novo mutations in ATP1A3 cause alternating hemiplegia of childhood. *Nature Genetics*, 2012; DOI: 10.1038/ng.2358

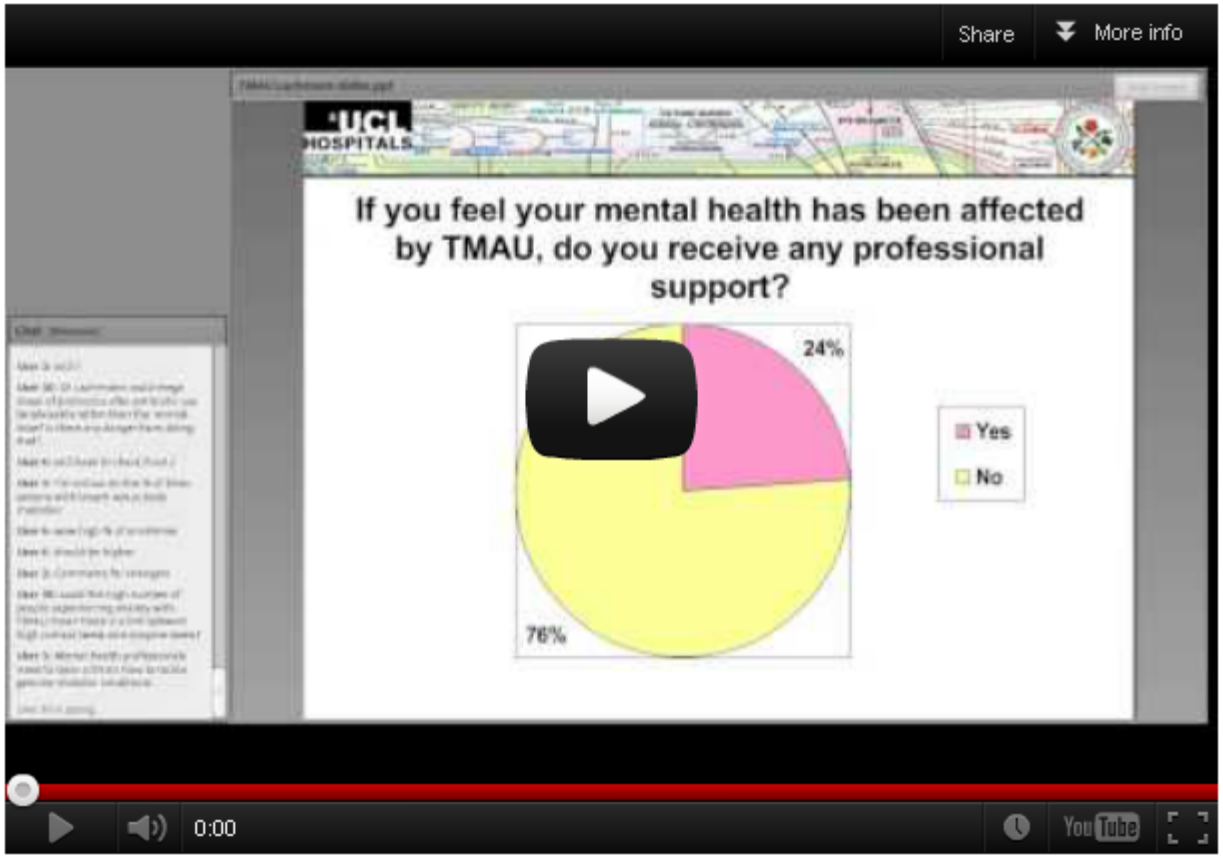
To view the free full text article, please click on the DOI numbers above.

The genetic cause of AHC discovered

July 29th, 2012 is so far, in the history of AHC research, the most important day. On this day, two groups of researchers, who have worked absolutely independently of each other, published the results of their work. The 1st publication was created by huge collaboration between research teams from several countries, the second one was produced by a German research team, on its' own. The first group's results were publicly received, at the end of April (through their advertised presentation, during the Congress of Child Neurologists in Brisbane). However, the 2nd publication was a complete surprise. Most importantly, the results of both groups are in agreement: De novo mutations in the gene ATP1A3 will cause, in most patients, the alternating hemiplegia of childhood.

3rd TMAU Webinar

Written by **rdc-team**, published 1 day ago.



This is the 3rd in a series of webinars which are a joint initiative betweeen RareConnect and TMAU advocates from the TMAU community.

For a number of years **Dr Robin Lachmann** has been the most prominent consultant for TMAU cases in the UK. Dr Lachmann is a Consultant in Metabolic Medicine in the Charles Dent Metabolic Unit in London. The unit helps and advises adolescents and adults with metabolic disorders.

Welcome - Alkaptonuria (AKU) Community

What is Alkaptonuria (AKU) ?

Alkaptonuria (AKU) is a rare autosomal recessive disease, affecting tyrosine metabolism. Patients accumulate homogentisic acid at 2000 times the normal rate, resulting in cartilage degradation and symptoms similar to early-onset osteoarthritis. **AKU Resources**

Latest Member Stories

More members stories

Charlotte's Story

by rdc-team published about 1 month ago
3 comments

'No I'm not AKU, no it wasn't an accident, but I don't really know why' I was born in 1963 with a missing left hand and a shortened forearm. My early years were filled with visits...

More

Monika's Story

by rdc-team published about 1 month ago
3 comments

My name is Monika. I was born in 1962 as a Contergan (AKU) baby in Germany with a short arm, a damaged hip and very weak shoulders. The fact that the hip was missing a bone ...

More

Partners and Patient Groups

RareConnect is a joint venture of Euordis and NORD. The following patient groups are official partners of this community.

- Rett Syndrome Europe
- Magyar Rett Szindróma Alapítvány
- Rett UK
- Asoc. Española del Síndrome de Rett
- Belgische Rett Syndroom Vereniging
- Asoc. Catalana del Síndrome de Rett
- Assoc. Nacional de Pais e Amigos Rett
- Rett Syndrome Poland
- Rett Syndrome Ireland
- Landsforeningen Rett Syndro
- Associazione Italiana Rett
- Norsk Forening for Rett Syndrom
- Österreichische Rett-Syndrom
- Rett Syndrom i Sverige
- Rett Syndrome South Africa
- Assoc. Française du Syndrome de Rett

More

Meet our members

View All

Latest AKU Community Activity

Allergy
topic, published about 1 month ago, show transcript

New abstract on AKU and shoulder prosthesis
topic, published 3 months ago
show transcript

Nitisinone
topic, published 3 months ago

New abstract on AKU and shoulder prosthesis
topic, published 3 months ago
show transcript

More

Alkaptonuria (AKU) Community

Learn how others live with Alkaptonuria (AKU) and share your story

SHARE YOUR STORY
(you must be signed in)

About Each Other

by Kracht, Dutch VHL Org. published 3 months ago

Anneke and Astrid from Holland talk about living with VHL.
Anneke van D. (52) has von Hippel-Lindau (VHL) disease, an inherited tumor syndrome in which multiple tumors in the body can occur. Her daughter, Astrid (15) recently heard that she does not have the disease.

Anneke:
"When I was thirteen, I lost my left eye. A serious

Read more

A life-saving hobby

by Greyowl55 published 3 months ago

How the internet and an enquiring mind are not necessarily dangerous! My life-long hobby has been genealogy; family history. Such things as ages and causes of death interest genealogists. I spotted a cluster of cancer related deaths at young ages in one distant section of the family, made contact and was told that they had VHL. Wondered why my father had died at 43 of a brain tumour. In 1965 these things were not known. Obtained his death certificate - a hemangioblastoma. Did some research, made the connection - approached my doctor with a...

Read more

Educating and Empowering People

by VHL Family Alliance published 8 months ago

An empowering message full of suggestions for activities.
I just returned from the 9th International Medical Symposium on von Hippel-Lindau, this year held in Rio de Janeiro, Brazil, hosted by the Brazilian National Cancer Institute (INCA) and chaired by Dr. Jose Claudio Casali da Rocha, head of the Brazilian National Tumor Bank, and co-chaired by Dr. Eric Jonasch, Genito-Urinary Oncologi...

Read more

Alkaptonuria (AKU) Community

Meet, discuss & support other patients or families living with Alkaptonuria (AKU). Contribute to topics, or just share what's on your mind

Start a discussion now (you must be signed in) START

View: Translations (EN) original language

Expand All

luna | published 2 months ago | Originally written in English

Allergy

I suffer since my childhood of different allergic reactions (rhinitis, cutaneous reactions and more recently I had twice a beginning of respiratory oedema without being able to determine exactly its origin). I'm wondering if there are other Aku- patients suffering from allergy?

I've been trying again to understand the causes of these reactions with different allergists with no specific result up to now.

REPORT Reply

robpleticha | Research | published 3 months ago | Originally written in English

AKU and shoulder prosthesis

Ochronotic arthropathy: diagnosis and management:
www.ncbi.nlm.nih.gov/pubmed/22482092?dopt=...
But what's a rritical review? critical review??

Reply

REPORT

luna | published 2 months ago | Originally written in English

Allergy

After these summer months, with a moody weather season and my health being as variable as the weather, here I am back to you taking a little time to write.

You certainly know that the AKU Society is organizing the 5th International Workshop on AKU This event will be held at the Medical Institution in Liverpool 18th-19th of November 2011.

REPORT

Reply

DenisAKU | published 3 months ago | Originally written in English

Yes, I do believe that it was meant to be 'critical' not 'rritical.' Even in scientific/medical publications, typos can occur. Especially translating from Greek to English. Here's another article by an Italian group: Musculoskelet Surg. 2012 Mar 24. Shoulder arthroplasty in alkaptonuric arthropathy: a clinical case report and literature review.

REPORT

Reply

robpleticha | Research | published 3 months ago | Originally written in English

5th International Workshop on AKU
A selection of presentations from those speakers who have given permission for their slideshows to be published are now available to download on the AKU

REPORT

Reply

Share This

Not a member? JOIN NOW

Search Community

I want to...

- Tell my story >
- Start a discussion >
- Meet other members >
- Learn about the disease >

Discussion Topics

- Diagnosis
- Treatment
- Research
- Finding a Doctor
- Living with AKu
- Family issues

View All

Members

ksaustria
Registered 19 days ago

mblay
Registered 24 days ago

Armayones
Registered about 1 month ago

BLACKSWAN_foundation
Registered about 1 month ago

marzennasadzak
Registered about 1 month ago

View all members

Latest AKU Community Activity

Allergy
topic, published about 1 month ago, show transcript

New abstract on AKU and shoulder prosthesis
topic, published 3 months ago
show transcript

Nitisinone
topic, published 3 months ago

New abstract on AKU and shoulder prosthesis
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Allergy
topic, published about 1 month ago, show transcript

More

Alkaptonuria (AKU) Community

Here's a selection of information from patients & professionals to better understand Alkaptonuria (AKU)

Here are some of the most frequently asked questions and their answers:

If you are a patient or carer with a question about living with AKU, about treatments, or other relevant information please email your question to FAQ@rarediseasecommunities.org and we will put it to the specialists who have agreed to answer your questions from time to time.

See all FAQ's

Will NTBC halt the production of HGA altogether or just reduce the amount produced?

NTBC blocks the enzyme that helps form HGA. Short-term studies have shown that the amount of HGA in the urine of patients with alkaptonuria is reduced by up to 95% when taking NTBC.

Tags: Treatments

Read full answer

When NTBC is given the OK, will it be accessible to everyone throughout the world or just certain countries?

This is certainly our goal, however this would also be up to the drug approval process of each country.

Tags: Treatments

Read full answer

Do males have bladder problems, other than the stones?

Several males have problems with urinary tract infections and urinary urgency and frequency probably related to obstruction from prostate stones.

Read full answer

Do all AKU sufferers suffer with raptured tendons, in legs, or is it just a freak occurrence?

Not all people with alkaptonuria have ruptured tendons, but they do occur often enough in AKU to be more that just a freak occurrence. The most frequent rupture site is the Achilles tendon. We have...

Read full answer

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topic, published about 1 month ago, show transcript


New abstract on AKU and shoulder prosthesis
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topic, published 3 months ago


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Nitisinone
topic, published 3 months ago


New abstract on AKU and shoulder prosthesis
topic, published 3 months ago
show transcript

More

RareConnect vs. Facebook?

- Human **translation**
- Moderator **support** and training
- Ongoing technical **improvements**
- **Non profit** model
- Allows for disease conversations and Facebook conversations to be **separate**
- 3 full time **Community Managers**
- Your conversations and stories are **there to stay**

Thanks, let us know how we can work together

More information:


www.facebook.com/rareconnect


@rareconnect

www.youtube.com/user/eurordis


robert.pleticha@eurordis.org
marta.campabadal@eurordis.org


+34 663 092 790