

WELCOME !

A community **by and **for**
people affected by
neuromuscular diseases**

Where do we come from ?

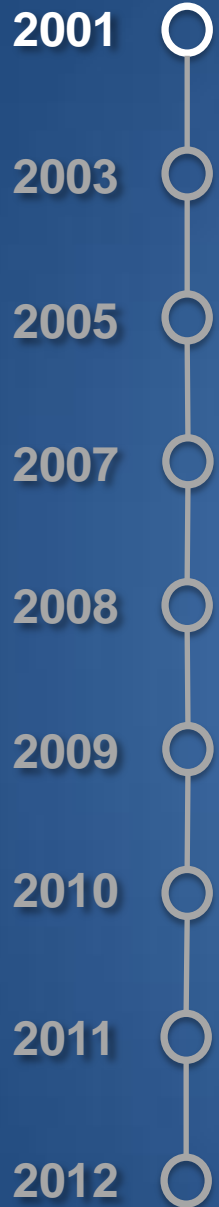
Where are we now ?

Where do we go ?

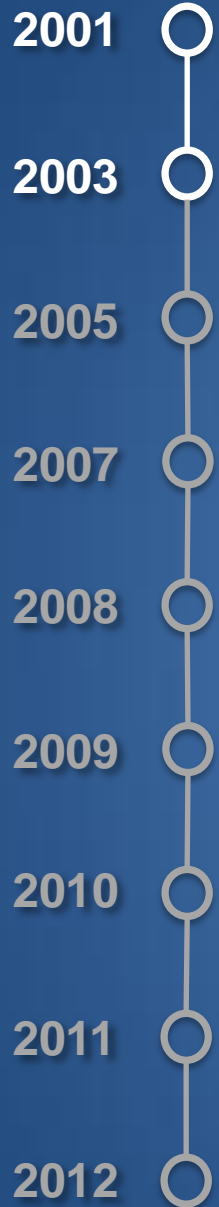
Where do we come from ?

Where are we now ?

Where do we go ?



A new diagnosis: Bethlem myopathy



**First scientific paper
describing the mechanism
of Bethlem myopathy**

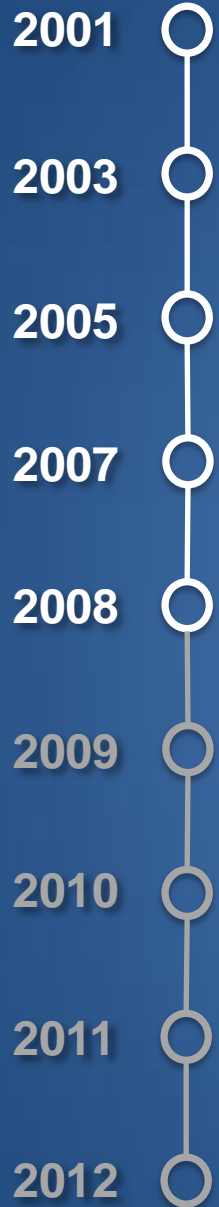


**Meeting with the first
other person affected
by the same disease**

2001 ○
2003 ○
2005 ○
2007 ○
2008 ○
2009 ○
2010 ○
2011 ○
2012 ○

**Meeting with the author
of the 2003 scientific paper**

Online publication of an interview



Launch of « Bethlem.info »

2001 ○
2003 ○
2005 ○
2007 ○
2008 ○
2009 ○
2010 ○
2011 ○
2012 ○

New non profit organization

CNIL Registration

**Launch of « Myopathies.info »
in French and English**



Volunteer based community



Hiring of Master students

**Content development
(articles, newsletter,
website improvements)**

Where do we come from ?

Where are we now ?

Where do we go ?

A community **by** and **for** people affected by neuromuscular diseases



**A website built on open
source technologies**

three simple words

FIND

MEET

SHARE

FIND

Interactive maps
Listing of patients
associations and
clinical centers

Scientific information
Scientists interviews

Accessible activities



Genetic transmission modes

Sunday, 10 April 2011 15:14 | Pauline NAUROY

Each cell of a human organism contains 46 chromosomes organized in 23 pairs, 22 coming from the father and 23 from the mother. Among these chromosomes, a pair determines the gender of a person: XX for women and XY for men. The 22 other pairs of chromosomes, non-sexual, are called autosomal.

Each chromosome has different information organized as genes, which determine different characters like the eyes color. Each gene can occur in different forms called alleles (for the example of the eyes color: blue, brown, green, etc.).

Chromosomes working by pairs, each cell has two copies of each-genes which can be either identical (we talk about homozygous. Example: blue/blue) or different (we talk about heterozygous. Example: blue/brown). When the two alleles of the same gene are different, one of the two alleles can dominate the other and determine the final character: we talk about "dominant". There's here a person "blue/brown" will have brown eyes. The "brown" allele is said to be "dominant" or the "blue" allele is then said to be "recessive".

NB: In all letters, a red person is a diseased person, a purple person is a healthy carrier but it don't express the disease and a blue person is a healthy person. In the same way a red dot is a mutated allele bearing the disease and a blue dot is a healthy allele.

AUTOSOMAL DOMINANT TRANSMISSION

A genetic disease is said to be transmitted in an autosomal dominant way if :

- The mutated allele is on a non sexual chromosome.

Interview of Paolo Bernardi

Sunday, 18 September 2011 20:39 | Pauline NAUROY

2008 Interview :

In 2003, Paolo Bernardi and Paolo Bonaldo published an article in Nature Genetics explaining the molecular mechanisms leading to muscle death in Bethlem and Ulrich myopathies after studying a mouse model of these diseases. Paolo Bernardi was kind enough to answer a few questions and we are glad to share with you his answers:

Myopathies.info : Could you summarize us your professional medical/scientific career?

Paolo Bernardi : I graduated in Medicine at the University of Padova, Italy, in 1976, where I also received postgraduate training in mitochondrial pathophysiology with Giovanni Fazio. I completed my scientific education with a postdoctoral stay in the laboratory of Harvey F. Lodish at the Whitehead Institute for Biomedical Research at MIT in Cambridge, Massachusetts. Back to Italy I served in the Faculty of Medicine of the University of Padova where I became Professor of General Pathology, and then Chair of the Department of Biomedical Sciences.

Myopathies.info : What lead you and your team to work on Bethlem/Ulrich muscular dystrophies?

I love ski !

Sunday, 27 February 2011 22:48 | Pauline NAUROY

Winter, cold, snow some words rhyming with skiing. Presentation of associations who permit you to ski everywhere.

The National Sports Center for the Disabled, in the United States

The National Sports Center for the Disabled was founded in 1970 to deliver ski lesson for children with amputations. Today the NSCD is one of the largest outdoor therapeutic recreation agencies in the world. It proposes lots of activities (like alpine skiing, kayaking, camping or horse pack trips) for people with disabilities (with Muscular Disorder, Muscular Dystrophy or Nerve Disorder for example). During the winter, the NSCD offers a variety of exciting adventures, including alpine skiing,

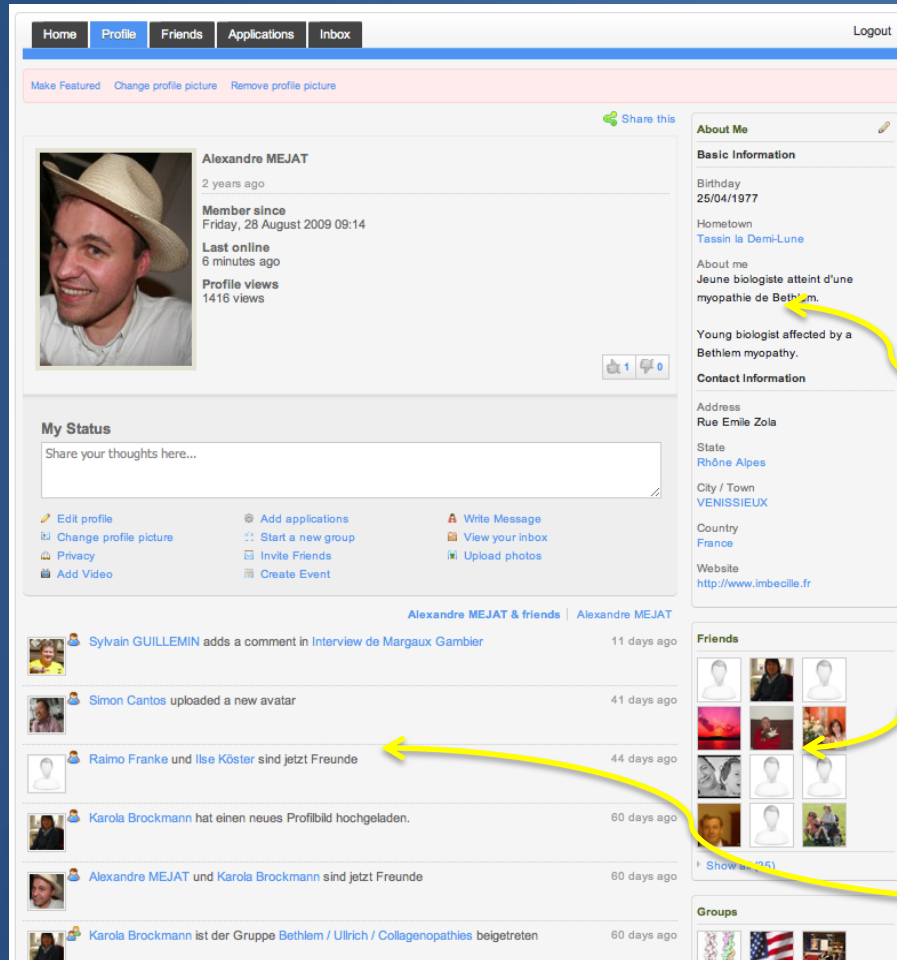
CONTACT INFORMATION TO SKI

United States of America :
<http://www.nscd.org/index.htm>

Canada :
<http://www.sportsadaptes.ca/fr>
<http://www.youtube.com>

France :
<http://appash.free.fr/>
<http://handimobility.org/blog/?p=6881>
<http://www.rhonealpes-tourisme.fr>

MEET



The screenshot shows a user profile for Alexandre MEJAT. The profile includes a navigation bar with 'Home', 'Profile', 'Friends', 'Applications', and 'Inbox'. Below the navigation bar are links for 'Make Featured', 'Change profile picture', and 'Remove profile picture'. The profile picture is a photo of a man wearing a hat. The profile information includes 'Member since Friday, 28 August 2009 09:14', 'Last online 6 minutes ago', and 'Profile views 1416 views'. The 'About Me' section contains 'Basic Information' such as 'Birthday 25/04/1977', 'Hometown Tassin la Demi-Lune', and 'About me Jeune biologiste atteint d'une myopathie de Bethlem.'. The 'Contact Information' section includes 'Address Rue Emile Zola', 'State Rhône Alpes', 'City / Town VENISSIEUX', 'Country France', and 'Website http://www.imbecille.fr'. The 'My Status' section has a text input field and buttons for 'Edit profile', 'Change profile picture', 'Add applications', 'Write Message', 'Start a new group', 'View your inbox', 'Privacy', 'Invite Friends', 'Upload photos', and 'Add Video', 'Create Event'. The 'Activity stream' section shows a list of recent activities, including 'Sylvain GUILLEMIN adds a comment in Interview de Margaux Gambier', 'Simon Cantos uploaded a new avatar', 'Raimo Franke und lise Köster sind jetzt Freunde', 'Karola Brockmann hat einen neues Profilbild hochgeladen.', 'Alexandre MEJAT und Karola Brockmann sind jetzt Freunde', and 'Karola Brockmann ist der Gruppe Bethlem / Ullrich / Collagenopathies beigetreten'. The 'Friends and groups' section shows a grid of profile pictures and a 'Show all (24)' link.

Personal information

Friends and groups

Activity stream

SHARE

Home Profile Friends Applications Inbox Logout

MENSCHEN MIT MUSKELKRANKHEIT OHNE DIAGNOSE (PRIVATE)

All Groups My Groups Pending Invitations Search Create

Report group Share this

Group Information

Category: Patients associations

Name: Menschen mit Muskelkrankheit ohne Diagnose (Private)

Created: Monday, 16 January 2012

Creator: Raimo Franke

Description

This is a German group for people with a myopathy without defined diagnosis.

Announcements

Willkommen

Sunday, 22 January 2012 by Raimo Franke

Herzlich willkommen bei der Gruppe "Menschen mit Muskelkrankheit ohne Diagnose"!

Diese Gruppe ist nur für Mitglieder sichtbar, d.h. aller hier veröffentlichten Beiträge sind nur für die Mitglieder bestimmt.

Hier ist ein guter Ort, um sich auszutauschen und näher kennen zu lernen. Bitte machen Sie Vorschläge, wie wir unsere Gruppe in Zukunft organisieren wollen, und welche Ideen Sie für die Gestaltung unserer Aktivitäten in der Zukunft haben.

Ich hoffe auf einen interessanten und regen Austausch!

Ihr
Raimo Franke

Displaying 1 of 1 bulletins

Discussions

Willkommen - Stellen Sie sich vor und teilen Sie Wünsche und Ideen für unsere Gruppe.

0 Replies

Started by Raimo Franke.

Group Option

- Edit Avatar
- Edit
- Send e-mail to members
- Create Bulletin
- Create Discussion
- Create Event
- Upload photos
- Create...
- Add Video
- Unpublish Group
- Join Group
- Delete Group

Admins

Show All (1)

Members

Show All (6)

Photo Albums

Displaying 1 of 1 albums

Show all albums

Group information

Administrator options

Discussions, events, Announcements, pictures and videos

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Where are we now ?

Where do we go ?

2001 ○
2003 ○
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2012 ○

Volunteer based translators group
(English, French, Italian, German, Spanish)

**New website with improved
interactivity and better control
on privacy**



muscleconnect.org
stronger together against myopathies

<http://www.myopathies.info>

MEET THE TEAM

myopathies.info
stronger together against myopathies



Alexandre
MEJAT
President



Patrice
BOUCHE
Treasurer



Pierre
BACHE-GABRIELSEN
Secretary

Thomas
COLLOT
Webmaster



Pauline
NAUROY
French - Editor



Donatella
RAGGI
Italian



Daniela
PERTIERRA
English and Spanish



Raimo
FRANKE
German

THANK YOU !