MYONEXUS THERAPEUTICS SECURES $2.5 MILLION SEED FINANCING TO CLINICALLY ADVANCE LIMB-GIRDLE MUSCULAR DYSTROPHY (LGMD) GENE THERAPIES

MYO-101 Phase 1/2a systemic gene therapy trial for LGMD2E to begin in early 2018.

NEW ALBANY, Ohio -- Myonexus Therapeutics, Inc. (Myonexus), a clinical-stage biotechnology company developing transformative gene therapies for limb-girdle muscular dystrophies (LGMDs), today announced the completion of a $2.5 million seed financing. The company is committed to accelerating development of five gene therapies pioneered within the lab of Louise Rodino-Klapac, Ph.D. and under clinical guidance by Jerry Mendell, M.D., each a Principal Investigator at Nationwide Children’s Hospital Center for Gene Therapy in Columbus, Ohio, USA. CincyTech, LLC, Rev1 Ventures, The Jain Foundation, and GFB ONLUS joined initial investors from the LGMD community.

The seed financing will enable Myonexus to initiate a systemic Phase 1/2a clinical trial of MYO-101, the company’s gene therapy candidate for treating LGMD2E (beta-sarcoglycanopathy), in early 2018. Myonexus’ pipeline also includes MYO-102, a gene therapy candidate for LGMD2D (alpha-sarcoglycanopathy) currently completing a Phase 1/2a
clinical trial, MYO-201, a gene therapy candidate for LGMD2B (dysferlinopathy) currently in Phase 1, MYO-103, a preclinical gene therapy candidate for LGMD2C (gamma-sarcoglycanopathy), and MYO-301, a preclinical gene therapy candidate for LGMD2L (anoctamin 5).

“MYO-101’s compelling preclinical data strongly supported the case for clinical translation, validated by our own subsequent clinical trial results as well as general advances in the neuromuscular disease gene therapy field,” said Michael Triplett, PhD, Myonexus’ President and Chief Executive Officer. “We are committed to rapidly advancing our pioneering pipeline of gene therapy candidates, with the ultimate goal of providing first-ever corrective LGMD treatments, and we plan to initiate the Phase 1/2a study of MYO-101 next year.”

“We have a history of investing in world class pediatric innovations in partnership with Cincinnati Children’s Hospital,” said John Rice, PhD, Director of Life Sciences at CincyTech. “We are pleased to find another investment with an immediate and lasting impact on children living with these challenging and rare diseases. Nationwide Children’s deserves great credit for developing this technology in house and proving it out clinically.”

“There are currently no approved treatments for the limb-girdle muscular dystrophies. Initial LGMD human clinical studies demonstrated expression in muscles exposed to MYO-102 and MYO-201. MYO-101’s Phase 1/2a clinical trial represents the first intravenous systemic exposure, potentially providing the first evidence of functional improvements in LGMD patients following treatment with these gene therapies,” said Bruce Halpryn, PhD, Myonexus’ Chief Operating Officer.

“With a ground-breaking approach Myonexus is poised to transform the way limb-girdle muscular dystrophy is treated, and we are thrilled to help accelerate their growth,” said Tom Walker, CEO of Rev1 Ventures. “This news underscores the critical role that the Research Institute at Nationwide Children’s Hospital is playing in advancing drug development research and identifying partners with the determination and ability to commercialize these discoveries for the ultimate benefit of patients. We believe the partnership with Myonexus is setting the stage for big things in gene therapy.”

“The Jain Foundation is excited to continue its support of the dysferlin gene therapy technologies emerging from Nationwide Children’s and be part of the clinical development
efforts happening at Myonexus. We believe gene therapy has great potential to be the first effective therapy for LGMD2B patients and to dramatically improve their quality of life,” said Doug Albrecht, PhD, Co-President of the Jain Foundation Inc.

About Myonexus Therapeutics

Myonexus Therapeutics is a clinical stage, rare disease gene therapy company developing first ever treatments for Limb-girdle muscular dystrophies (LGMDs) based on research at Nationwide Children’s Hospital, a leader in neuromuscular gene therapy discovery and translational research. Myonexus Therapeutics’ pipeline includes three clinical stage gene therapy programs (LGMD2E, LGMD2D, and LGMD2B) and two preclinical gene therapy programs (LGMD2C and LGMD2LO). Founded in 2017, Myonexus is headquartered in New Albany, Ohio. More information is available at www.myonexustx.com.

About CincyTech LLC

CincyTech, one of the Midwest's most active seed investors, helps to transform innovation into high performing life science and digital companies in Southwest Ohio. Our team provides advice and seed capital to entrepreneurs, helps research institutions commercialize technology through startups, and catalyzes investment from individuals and institutions into regional companies. Learn more at http://cincytechusa.com.

About Rev1 Ventures

Rev1 is a venture fund that helps entrepreneurs build great companies. Combining investment capital with a unique blend of services through our startup studio, we propel innovation for startups and corporate innovation teams. Our seasoned, data-driven team helps lay the foundation for scalable growth with the skills to evolve a product, sell to customers, and build the right team. Named a top VC investor in the Great Lakes Region, Rev1 manages a continuum of financial support from corporate and community partners, as well as the Ohio Third Frontier. Rev1 was named the Most Active VC in Ohio in 2017 by CB Insights. For more information, visit http://www.rev1ventures.com.
About the Jain Foundation

The Jain Foundation is a privately funded nonprofit foundation focused on finding a cure for muscular dystrophies caused by dysferlin deficiency (LGMD2B/Miyoshi Myopathy). The Jain Foundation supports academic research, conducts independent studies with contract research organizations, and supports clinical studies in pursuit of this mission. The foundation also assists patients in receiving a confirmed genetic diagnosis of dysferlinopathy, and maintains a patient registry for the disease.

PUBLISHED IMPORTANT RESULTS FOR SMA1 TRIAL

On November 2\textsuperscript{nd} the magazine The New England Journal of Medicine published the article “Single-Dose Gene-Replacement Therapy for Spinal Muscular Atrophy”, the authors are K. Kaspar e J. Mendell. In the article there are all the results of the SMA1 trial of gene therapy.

See the article at the link: \url{http://www.nejm.org/doi/full/10.1056/NEJMoa1706198#.WfsctKM_52d.facebook}

Spinal muscular atrophy type 1 (SMA1) is a progressive, monogenic motor neuron disease with an onset during infancy that results in failure to achieve motor milestones and in death or the
need for mechanical ventilation by 2 years of age. They studied functional replacement of the mutated gene encoding survival motor neuron 1 (SMN1) in this disease.

Fifteen patients with SMA1 received a single dose of intravenous adeno-associated virus serotype 9 carrying SMN complementary DNA encoding the missing SMN protein. Three of the patients received a low dose, and 12 received a high dose. The primary outcome was safety. The secondary outcome was the time until death or the need for permanent ventilatory assistance.

As of the data cutoff on August 7, 2017, all 15 patients were alive and event-free at 20 months of age, as compared with a rate of survival of 8% in a historical cohort. Of the 12 patients who had received the high dose, 11 sat unassisted, 9 rolled over, 11 fed orally and could speak, and 2 walked independently.

In patients with SMA1, a single intravenous infusion of adeno-associated viral vector containing DNA coding for SMN resulted in longer survival, superior achievement of motor milestones, and better motor function than in historical cohorts. Further studies are necessary to confirm the safety and efficacy of this gene therapy.

FDA CLEARS IND APPLICATION FOR MICRO-DYSTROPHIN
GENE THERAPY PROGRAM TO TREAT DUCHENNE

The U.S. Food and Drug Administration (FDA) has cleared an investigational new drug application for a micro-dystrophin gene therapy program by Sarepta Therapeutics and Nationwide Children’s Hospital, in clinical development for Duchenne muscular dystrophy (DMD).

A Phase 1/2a clinical trial is now enrolling participants and should begin dosing later in December.

The study will assess the safety and tolerability of AAVrh74.MHCK7.micro-Dystrophin in DMD patients. The trial is also designed to evaluate biological activity and efficacy of the micro-dystrophin vector by seeing how it performs when replacing the missing muscle dystrophin in DMD.

Between 60 to 70 percent of Duchenne boys could be potential candidates for this treatment,
since researchers are looking for patients with mutations between exons 18 and 58.

For the trial, 12 patients will be split into two groups to undergo gene transfer and establish maximum tolerated dose (MTD) avoiding toxicity. One group will include infants three months to three years of age with DMD; the second will boys 4 to 7 years old.

The principal investigators, Dr. Jerry Mendell and Louise Rodino-Klapac of Nationwide Children’s in Columbus, Ohio, will conduct the Phase 1/2a study. It'll use a construct (MHCK7) developed by Mendell and Rodino-Klapac to enhance activity of the micro-dystrophin vector in DMD patients. The construct can be delivered to skeletal, diaphragm and cardiac muscles and has shown high levels of gene expression in preclinical studies. Micro-dystrophin expression will be measured at three months, via biopsy.

This is the first micro-dystrophin gene therapy program in development for DMD. If approved, Sarepta has the option to retain worldwide exclusive rights.

“This will be the first clinical trial for DMD to treat patients as young as three months old, and is an important step forward in our quest to diagnose patients through newborn screenings,” Mendell, who also heads the neuromuscular gene therapy program at Nationwide, said in a press release. “We are very encouraged by the promising preclinical results and are eager to complete enrollment.”
Added Rodino-Klapac, principal investigator at Nationwide’s Center for Gene Therapy: “With this clinical trial, we are laser-focused on giving each patient the best possible chance of a successful outcome. We are taking a novel approach to trial design. The use of potentially therapeutic doses is a critical part of our mission to arm our patients for their one shot at gene therapy.”

The clinical trial received $2.2 million in funding from Parent Project Muscular Dystrophy (PPMD). Recently, Sarepta and Nationwide announced the clearance of another IND application, regarding its GALGT2 gene therapy program for DMD.

“As the first systemic micro-dystrophin gene therapy program enters the clinic,” said Sarepta President and CEO Douglas Ingram, “we mark an important milestone in our journey to relentlessly pursue new therapies to treat DMD.”
GFB ONLUS STARTS DREAMING AGAIN,

NOW MUSCULAR DYSTROPHY HAS THE COUNTED DAYS:

Within 2027 there will be five new therapies

Great emotion at GFB Onlus, that has reached an important achievement in scientific research for curing the limb-girdle muscular dystrophy, a rare form of dystrophy that in a short time does not permit to walk anymore, then to breath and then it stops the heart.

On June 15th our Onlus signed an important agreement with an American Company - Myoexus Therapeutics – with the objective of achieving, within 2027, the acknowledgment of five new specific therapies for different forms of limb-girdle muscular dystrophies.

With this agreements GFB Onlus has officially been admitted to the said Company and has sent to Columbus other 200.000 Euros for supporting the project.

Starting from 2012 this project - carried out by Professor Jerry Mendell at the Nationwide Children’s Hospital of Columbus, Ohio, in the United States – has exclusively been financed by GFB Onlus, with a total amount of 1.322.500 dollars.

The results obtained in these years are in two scientific publications and the pre-clinical phase
News

has been completed thanks to the great commitment of GFB Onlus, only financing body. «Just last year, but, GFB Onlus was afraid not to be able to continue alone with the development of this therapy – says Beatrice Vola, chairman of GFB Onlus – the future appeared to be uncertain … but this year there was an important change and after a year we had to stop because of the lack of funds, finally now the American project of genetic therapy can restart». It begins so a “new era” for GFB Onlus, that is not alone anymore to finance this project: other groups of American families have joined the Company. They have a similar story to GFB, to underline that what is in Italy it is also in other countries, where the resources seem to be more. These families’ children suffer from an unknown illness, there are no bodies financing the researches and the families feel to be forgotten. So they decide to create foundations on their own, that operate in this field.

Now these foundations are joining to GFB Onlus in the Company Myonexus and all together they be stronger!

The Company has been active in founding more and more funders and already an important American company has been involved, this is REV1 Ventures.

Moreover other bodies are getting in touch with the Executive Director of the company, Dottor Michael Triplett. For further info see link www.myonexustx.com.

“the programme of the company is very ambitious, within 2027 there will be five new therapies for some forms of limb-girdle muscular dystrophies (LGMD2B-2C-2D-2E-2L) – says Mrs Vola enthusiastic – This year the trial on patients with 2E will start, and that possible thanks to the project financed by GFB ONLUS, to pass then in the next years to the other four diseases. There will be injected the first 6 patients by systemic route, at dosages much higher.

The therapy will reach the whole body, heart too. In 2017 almost 2.000.000 dollars will be spent on trial with 2E, that will involve much more patients”. 
GFB is now working actively to find other financiers for the Company and to reach another important goal: to take the therapy also to Milan in few years – in 2020. With these goals, GFB Onlus has started the campaign “Let’s treat them 4.0” for improving patients’ quality of life suffering from limb-girdle muscular dystrophy.

“All this seems to be still a dream for the members of the association – says Beatrice Vola – but it was possible thanks to the many supporters, in particular to all those that participated in the loan “Terzo Valore” and to all those that supported the first campaign SMS of GFB in May.

Thanks to the collected funds we now think to be already able to send a further transfer to the Company next year.

The association thanks its friends Luca Ciaponi, the families of Roma, Ancona, Viareggio, Lecco, Como, Sondrio, a French family and the bodies that participated, Cooperativa Orizzonte, Proloco of Paniga, group Presepe Cà Giovanni, the theatre company Amici Anziani, Tecnici Senza Barriere, our friends skiers, Nuovo Pignone of Talamona, the association Amici del Bambino, the sections Uildm of Sondrio and Lecco, the working group Association Amici Anziani ».

All further info at www.lgmd2e.org

PUBLISHED THE SECOND PHASE OF THE AMERICAN PROJECT OF GFB ONLUS
On April 5th the magazine Molecular Therapy published the article “Systemic AAV-Mediated Sarcoglycan Delivery Targeting Cardiac and Skeletal Muscle Ameliorates Histological and Functional Deficits in LGMD2E Mice”. In the article are all the results of the second part of the pre-clinical phase of the project of gene therapy financed by GFB ONLUS.

See the article at the link: https://www.ncbi.nlm.nih.gov/pubmed/28284983

OTHER PATIENTS AFFECTED FROM LGMD2C-2D-2E IN GFB ONLUS

In the last months GFB ONLUS has found other patients affected from Lgmd2c-2d-2e. GFB counts now a total of 203 patients affected from Sarcoglycanopathy, so divided:

LGMD2C

LGMD2D
LGMD2E

LGMD2F

SARCOGL.

2010

0

1

5

0

2013

4
On the website of the association you can find the list of the patients sorted by geographical provenance to the link:

http://www.beta-sarcoglicanopatie.it/index.php?option=com_content&view=article&id=46&Itemid=54
GFB ONLUS HAS PARTICIPATED IN ENMC WORKSHOP

In March Prof. Yvan Torrente represented GFB ONLUS at the Workshop organised by ENMC (European Neuro Muscular Centre), held in Narden, in Netherlands. Title of the workshop “Limb Girdle Muscular Dystrophies, Nomenclature and reformed Classification”. During the workshop it was introduced a new nomenclature for the Limb Girdle Muscular Dystrophies.


GFB ONLUS HAS PARTICIPATED IN THE CONVENTION TELETHON

Dr. Carles Sanchez Riera, member of the Medical and Scientific Commission represented GFB ONLUS at the Convention Telethon, held in Riva del Garda, Italy, in March. He participated at the convention of the Associazioni Amiche of Telethon and at the meeting of Coordinamento Associazioni Malattie Neuromuscolari CAMN (Coordination Associations for rare neuromuscular diseases)
GFB ONLUS HAS RECEIVED OTHER CONTRIBUTIONS

The GFB Onlus has received the following contributions:

2017:

__82.435

Quote prestito Terzo Valore,

Proloco di Paniga, Tecnici Senza Barriere, sezione UILDM di Lecco, Amici Sciatori e 27 privati

51.565 €

Donazioni Terzo Valore,

amici di Luca Ciaponi, Cooperativa Orizzonte, compagnia teatrale Amici Anziani, gruppo Presepe Cà Gi
News

65.600 €

Prestito Banca Prossima

18.526 €

Biglietti lotteria GFB

225 €

Tessere sostenitori GFB

9.872 €

Contributo 5x1000 relativo al 2015

50 €

Donazione NN per la ricerca scientifica
100 €

Donazione NN per la ricerca scientifica

20 €

Donazione NN per la ricerca scientifica

150 €

Donazione Coscritti 1941 di Talamona

30 €

Donazione NN per la ricerca scientifica

50 €

Donazione NN per la ricerca scientifica
100 €

Donazione NN per la ricerca scientifica

1.200 €

Donazione NN per la ricerca scientifica dalla Francia

100 €

Donazione NN per la ricerca scientifica

500 €

Donazione NN per la ricerca scientifica

200 €

Coscritti 1949 di Talamona in memoria di Vito

500 €
Gruppo di lavoro Associazione Amici Anziani Talamona

1.300 €

Donazione Nuovo Pignone GE

5.973,09 €

Bando Home Care Premium

20 €

Donazione NN per la ricerca scientifica

250 €

Donazione NN per la ricerca scientifica

11.530 €
Convenzioni con Enti Pubblici

500 €

Donazione NN per la ricerca scientifica

20 €

Donazione NN per la ricerca scientifica

50 €

Donazione NN per la ricerca scientifica

6.738 €

Campagna SMS maggio [link a campagna SMS]
15 €

Donazione NN per la ricerca scientifica

10 €

Donazione NN per la ricerca scientifica

879 €

Donazione Torneo Cral Nuovo Pignone per la ricerca scientifica

50 €

Donazione NN per la ricerca scientifica

20 €

Donazione NN per la ricerca scientifica
25 €

Donazione NN per la ricerca scientifica

4.000 €

Donazione NUOVO PIGNONE SRL per la ricerca scientifica

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**SUPPORT US**

Any amount you want donate for the research will be a real help in the fight against the beta-sarcoglycanopathies and the other forms of muscular dystrophy.
FUND FOR RESEARCH:

For this purpose a special American projects.

HEADING FOR RESEARCH FUND: Gruppo Familiari Beta-sarcoglicanopatie Onlus

IBAN: IT33X0335901600100000076500

BIC /SWIFT code BCITITMX

PAYPAL TRANSFER

HEADING: Gruppo Familiari Beta-sarcoglicanopatie Onlus

EMAIL: fo@beta-sarcoglicanopatie.it
GFB ONLUS HAS SENT OTHER 200.000 $ TO COLUMBUS

Another big effort for the association with its offices in Talamona in the province of Sondrio. On Friday, 9th September GFB ONLUS sent another contribution to Columbus Ohio, of 202.500 $. From 2012 up to now we have financially supported the American laboratories with a total of 1.300.000 $. This further contribution will permit Prof. Mendell to go on with the project of GENE THERAPY FOR LGMD2E. GFB is at the moment the only financier of the project and without this support the project is destined to stop. The funds sent will be necessary for preparing the systemic clinical trial in high doses, that should be carried out in 2017.

This amount now sent will not be sufficient to make the clinical trial re-start. It is foreseen a further bank transfer of the association to Columbus within the end of the year.

You will find all the info about the project to the following link:

http://www.beta-sarcoglicanopatie.it/index.php?option=com_content&view=article&id=166&Itemid=111
GENE THERAPY IS GOING TO BECOME A DRUG

The gene therapy injected in patients affected by SMA1 has given very good improvements in the first 15 children treated in Columbus Ohio. Thanks to these results it will be possible to develop an abbreviated procedure to make this therapy become a drug. This procedure can be used since the therapy has been developed in order to treat a serious disease that is dangerous for the life and permits an important improvement.

This makes us hope also for the use of this therapy for our LGMD2E.


HELP US TO FINANCE THE THERAPY

GFB ONLUS intends to continue also in the future to support this project in Columbus. For supporting the project of gene therapy for LGMD2E, GFB ONLUS has created a special FUND FOR RESEARCH through its current account of Banca Prossima. All the donations received on that account will be used to support the American projects and the scientific research on LGMD2E. Be one of us, and give your support with a donation on the bank account:

**HEADING FONDO PER LA RICERCA:** Gruppo Familiari Beta-sarcoglicanopatie Onlus

**IBAN:** IT33X0335901600100000076500

**BIC/SWIFT code:** BCITITMX
GFB ONLUS ASKS FOR SUPPORT TO BANKS

THE FUNDS COLLECTED IN THESE YEARS ARE NOT SUFFICIENT TO SUPPORT THE PROJECTS OF THE ASSOCIATION, therefore starting from now GFB ONLUS is addressing to banks and is getting some loans.

The first loan already requested is “SUBITO 5X1000”, through which our association has already received a contribution of 5x1000 of this year of 8.048,46 €.

Moreover we are informing for getting other loans, among which “Terzo Valore”, whose news will be given in the next newsletters.
OTHER PATIENTS AFFECTED FROM LGMD2C IN GFB ONLUS

In the last months GFB ONLUS has found 5 other patients affected from Lgmd2c-2d-2e. These patients are from Tunisia, the Philippines, United States and Italy (Pescara). GFB counts now a total of 126 patients affected from Sarcoglycanopathy, so divided:

<table>
<thead>
<tr>
<th>LGMD2C</th>
<th>LGMD2D</th>
<th>LGMD2E</th>
<th>LGMD2F</th>
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</thead>
<tbody>
<tr>
<td>0</td>
<td>30</td>
<td>85</td>
<td>0</td>
</tr>
</tbody>
</table>

2010 1
On the website of the association you can find the list of the patients sorted by geographical provenance to the link:

http://www.beta-sarcoglicanopatie.it/index.php?option=com_content&view=article&id=46&Itemid=54

ALESSIA: A NEW LIFE THANKS TO JARVIK 2000

Great emotion among all the members of GFB ONLUS and a great sigh of relief for Alessia's story, that in the month of September overcame a delicate surgical operation, with implantation
of the device Jarvik 2000 in the heart.

Alessia’s story is present and readable at the link below:


Various stories of some other patients are also on our site at the link:


and if you also want to add your story on the site, mail your material to:

info@beta-sarcoglicanopatie.it
MEETING OF GFB ONLUS IN LYONS WITH PROF. MENDELL

On March, 17th GFB ONLUS took part to the congress *Myology 2016* in Lyons, where we met Prof. Mendell.

Present were Beatrice Vola (President of GFB), Marco Perlini (Vice President), Dr. Francesca De Santis (Polyclinic Hospital of Milan) and an interpreter.

During the meeting Prof. Mendell informed us about the important results obtained on SMA1 clinical trial. These will allow the project on LGMD2E now financed by GFB to make a great leap forward. The clinical trial foreseen in a muscle of a finger (intramuscular) will not be done, but it will be treated by injecting the therapy throughout the body of six-ambulatory patients (systemic).

In the next months the project will continue with the production of the new vector and the demand for the necessary authorizations for injecting the therapy in the first patients.
GFB ONLUS WILL TAKE PART TO THE WORKSHOP ENMC

Already three members of the Medical and Scientific Commission of GFB ONLUS have given their availability to take part to the Workshop ENMC with the title “Limb Girdle Muscular Dystrophies - Nomenclature and reformed Classification”. In winter some of the main experts of Limb Girdle Muscular Dystrophies will meet in Holland for three days. Here 21 participants will be present, among them there will be 4 associations of patients: Jain Fundation, Lgmd2iFund, Muscular Dystrophy United Kingdom and GFB ONLUS.

CARLES SANCHEZ: NEW MEMBER OF MEDICAL AND SCIENTIFIC COMMISSION OF GFB ONLUS

From the month of March the Medical and Scientific Commission of GFB ONLUS was strengthened with the arrival of Dr. Carles Sanchez, Spanish, that is now working in Rome with Prof. Pier Lorenzo Puri, in the labs of the Foundation Santa Lucia (Rome, Italy). The Commission is currently composed of 5 members: Prof. Massimiliano Cerletti, Roberto Maggi and Yvan Torrente, Dr. Paola Bonetti, Carles Sanchez.
OTHER PATIENTS WITH LGMD2C-2D-2E IN GFB ONLUS

In the last months GFB ONLUS has found other 14 patients with Lgmd2c-2d-2e. Three of them are Italian, three are from Libya, Palestine, Germany and the rest from the United States. In total 109 patients with Sarcoglycanopathy are now present in GFB, and they are so divided:

LGMD2C
LGMD2D
LGMD2E
On the site of the association is present the list of the patients sorted by geographical origin at the link:

http://www.beta-sarcoglicanopatie.it/index.php?option=com_content&view=article&id=46&Itemid=54
BALANCE SHEET 2015 OF GFB ONLUS APPROVED

On April, 15th it was approved the balance sheet 2015 of the association, this is the third balance sheet of the association, that has quadrupled the balance sheet of the previous year, with a final total of 323.562,12 €.

Thanks really very much to all those that have supported us and allowed us to continue with our projects. All together we can!

GFB ONLUS HAS FINANCED THE FIRST CLINICAL TRIAL FOR THE LGMD2E

On 20th November it was held the fifth conference call with Dr. Jerry Mendell and Dr. Rodino Klapac Louise and Dr. Erik Pozsgai Some families of the GFB ONLUS and Dr. Massimiliano Cerletti, a member of the Medical-Scientific Committee of the Association were also present
during the call. Discussion topics were the reports with all the results of the preclinical phase sent by Dr. Mendell in November and the various authorizations necessary for the first clinical trial for the LGMD2E. Dr. Mendell also communicated us the vector was modified by inserting a promoter specific for heart and diaphragm so to realise a therapy also helpful for treating the heart and breathing.

In the American laboratories they are already working to get ready for the systemic injection in the whole body of the patients.

After considering the positive results of the reports, GFB ONLUS has decided to go on financing the project and we have sent a bank transfer of 200,000$ to Columbus for starting already for January with the clinical trial, that will be on 3 patients over 18 years old. Foreseen for January a second amount of 150,000$ for the conclusion of the first trial that will last one year. GFB ONLUS is willing to support the project in Columbus also in the future.

For financing these researches GFB ONLUS has created a special FUND FOR THE RESEARCH with its bank account of Banca Prossima.

All the donations received on this bank account will be used for financing the American projects and the scientific research on the LGMD2E.

FUND FOR THE RESEARCH: Gruppo Familiari Beta-sarcoglicanopatie Onlus

Code IBAN: IT33X0335901600100000076500 BIC/SWIFT code BCITITMX

All the information on the project and updated information will be present on the site of the Association:
VOTE THE VIDEO OF GFB ONLUS IN THE CONTEST 1° NATIONAL PRIZE
BARRIER-FREE TOURISM

Until the 22nd February 2016 you can vote the video of GFB ONLUS on the event: “GFB ONLUS celebrates the arrival of its bus”, on the site Premio turismi accessibili (Prize Barrier-free tourism).

Before accessing the link it is necessary a registration in the field “registrati” (Registration) and then you can vote in the field “Vota” (Vote)

http://www.turismipertutti.it/articoli/item/38-il-gfb-onlus-festeggia-l-arrivo-del-suo-pulmino#add-review
THIRD ANNUAL MEETING OF GFB ONLUS

Also in 2016 GFB ONLUS proposes the third annual meeting of the families of the Association and supporters, that will be held from 13th to 16th August in Valmalenco (SO - Italy). This meeting is part of the project “Sport, environment and disability” of the Association, financed by the foundation Provaltellina, with the call “Services to the Person”.

It includes a helicopter flight and a boat cruise on Como lake. Anyone interested in taking part, can enroll by sending an e-mail to info@beta-sarcoglicanopatie.it
CONSORTIUM OF LGMD FOUNDATIONS ANNOUNCE FIRST ANNUAL LGMD AWARENESS DAY - 30 SEPTEMBER 2015

The first annual Limb Girdle Muscular Dystrophy (LGMD) Awareness Day is going to be launched later this year thanks to the unified effort of a number of non-profit foundations from around the world that are collaborating to promote global awareness.

For learning more about what is going to happen in September please visit lgmd-info.org. The site also features spotlight interviews and LGMD resources along with information about patient organizations.

If you are interested in participating in the first ever LGMD Awareness Day, there is a number of things you can do. These include becoming an "Ambassador" for LGMD Awareness Day or organizing an event to raise awareness and (if interested) solicit donations for your favourite LGMD charity.

To find out more about what others are doing please visit the Limb Girdle Awareness Day Facebook page
KINECT, A VIDEOGAME FOR CLINICAL TRIALS

A team at the National Children Hospital of Columbus Ohio believes that the videogame KINECT could be used in the future clinical trials for all those patients with serious muscular deficits.


A CLINICAL STUDY ON ABOUT THIRTY PATIENTS WITH LGMD2E WAS PUBLISHED

On 28th April 2015 the magazine “American Academy of Neurology” published a clinical study on the LGMD2E carried out by E. Pegoraro and C. Semplicini with some other European centres. The clinical study was carried out on thirty-two patients followed in various European clinical centres. In the article are many information about motor, respiratory and cardiac problems of patients. Some families of GFB ONLUS have been included in this study.
SECOND MEETING IN NAPLES OF THE LGMD EuroNET

On Friday 22nd May 2015 LGMD EuroNET had its second meeting in Naples during the 15° National Congress AIM. In the section “Future Projects and Programs”, Vola Beatrice (president of GFB ONLUS) and Prof. Corrado Angelini presented the LGMD EuroNET, followed by a meeting of the LGMD EuroNET, for planning the request for an European project for girdle muscular dystrophies. GFB ONLUS also showed the poster titled “Beta-sarcoglycanopathy: what’s new?”

More information: www.beta-sarcoglicanopatie.it
THE REPORTS OF THE MEETING “LGMD DAYS”, ORGANISED BY GFB ONLUS, IN THE MAGAZINE ACTA MYOLOGICA

In the month of December 2014 the magazine ACTA MYOLOGICA published the reports of the meeting “LGMD DAYS”, organised by GFB ONLUS in Lido di Venezia, in the days 15-17 October 2014.

http://www.ncbi.nlm.nih.gov/pmc/articles/PMC4369845/
GFB ONLUS HAS BEEN INCLUDED IN THE WEBSITE OF THE TREAT-NMD

The presentation of GFB ONLUS has been included in the website of the TREAT-NMD, in the section dedicated to the patient organizations for the girdle muscular dystrophies, to the link

http://www.treat-nmd.eu/lgmd/patient-organizations/

MARCO GAVAZZI, AMBASSADOR OF GFB ONLUS

Marco Gavazzi, left in May 18th

He walked for about 40 kms a day, for 25 days, starting from SENTIERO DEL VIANDANTE, up to Lecco...
SECOND ANNUAL MEETING OF GFB ONLUS

GFB ONLUS has organized its Second Annual Meeting, that was held in Valmalenco (Italy) in August from Saturday 8th to Thursday 11th.

On Sunday 9th it was planned a trip by the Red Train of Bernina, from Tirano (Italy) to St. Moritz (Switzerland).

In August, on Monday 10th, the families of GFB ONLUS have planned to go to Lake Palù in Chiesa Valmalenco.

More information to the link : info@beta-sarcoglicanopatie.it
A NEW BUS FOR GFB ONLUS

In July, on Saturday 11th, GFB ONLUS inaugurated its new bus equipped for the transport for disabled people, bought thanks to the contribution of numerous sponsors (Fondazione Provaltellina, Società Iperal Spa, BIM, Comunità Montana di Morbegno, Coro Onevoice di Roma, ditta Comtech, Fondazione Creval, Uildm Sondrio, Banca Intesa S. Paolo).

http://www.valtellinanews.it/articoli/Talamona-il-GFB-Onlus-festeggia-l-arrivo-del-nuovo-pulmino-FOTO-20150712/

https://www.youtube.com/watch?v=pqPgZgh8
THEATRE IN TREVISO IN SUPPORT OF GFB ONLUS

In June, on Saturday 27th a theatre was held in Treviso (Italy) in support of GFB ONLUS, organised by some families of patients living in the area. Anyone who wants to support our Association can organise a fundraising event in his/her town/city by contacting the secretariat of GFB ONLUS number +393497244391 or mailing to info@beta-sarcoglicanopatie.it

A NEW PATIENT SUFFERING FROM LGMD2E IS NOW PART OF GFB ONLUS FROM NORTH CAROLINA USA

From the month of June a new American family, living in North Carolina, is part of GFB ONLUS. The association has at the moment 27 patients suffering from LGMD2E and other 59 patients suffering from another sarcoglycanopathy.
IRCCS E. MEDEA: MEETING "LIMB GIRDLE MUSCULAR DYSTROPHY FROM DEFICIT OF CALPAIN 3"

Save the date: the Meeting, organised in cooperation with Associazione Italiana Calpaina 3 onlus (AlCa3) and Policlinico di Milano, thanks to interventions of national and international experts, is going to be held in November 14th 2015 at the Scientific Institute E. Medea a Bosisio Parini (LC). It is intended to provide patients with an updated picture of the state of the art in the diagnosis and prospects of therapy and rehabilitation of girdle dystrophy. Read the program at:

FUNDED THE PROJECT OF DORIANNA SANDONA’ ON THE LIMB GIRDLE MUSCULAR DYSTROPHY

The Italian foundation Telethon has funded the project of Dorianna Sandonà, that will test the efficacy of a new potential drug therapy in the animal model of sarcoglycanopathy.


THE GFB ONLUS AND PROF. ANGELINI CORRADO HAS ORGANIZED THE CONGRESS "LGMD DAYS"

On days 15, 16, 17 October will be held the Conference "LGMD DAYS" in Lido di Venezia (VE)
at the Institute IRCCS "San Camillo," Via Alberoni no. 70.

The Conference, in Italian language, will be entirely devoted to the clinical aspects of Limb Girdle Muscular Dystrophy, with a special focus on Sarcoglycanopathies.

On the Association's website, at the following link, have been published

- The provisional program of the conference,

- the registration form,

- any accommodation in Hotel,

- information on ferries to reach the location of the congress.


THE MYO-SEQ PROJECT: THE CTSR MADE IT POSSIBLE
There are a number of specific challenges at every stage of rare disease (RD) therapy development: incomplete knowledge and understanding of disease prevalence and disease course, genotype-phenotype correlation, modifying factors, response to treatment, potential side-effects, as well as difficulties in accessing biomaterials and identifying patients. In the neuromuscular field, that last specific challenge has become less cumbersome thanks to the existence of the Care and Trial Site Registry (CTSR).

About MYO-SEQ: Next Generation Sequencing (NGS) and other -omics technologies, in combination with powerful IT infrastructure and sophisticated bioinformatics tools, allows, in principle, the entire genetic profile (exome, genome) of any RD patient to be deciphered. It can also enable the integration of this information with clinical data (e.g. from deep phenotyping, ontologies, electronic health records) as well as data from clinical studies (such as response to treatment, side effects, pharmacogenomics) and biomarker studies (proteomics, metabolomics).

In this context, the neuromuscular team at Newcastle University has conceived the MYO-SEQ project that will focus on the application of NGS, in particular Whole Exome Sequencing (WES), in a large cohort of patients with unexplained limb-girdle weakness (LGW). Focusing on undiagnosed patients with a clearly defined clinical phenotype will enable increased diagnostic rates for disease causing mutations in known genes in this cohort, while the use of WES provides scope both for new gene discovery and for additional research into disease modifiers and genotype-phenotype correlation with substantial cost effectiveness.

The initial inquiry to test the feasibility of the study was carried out with a short questionnaire publicized via the CTSR. The excellent response from the CTSR affiliated clinical centres has directly resulted in the engagement of private and non-for-profit funders to support the execution of the project that is expected to start in August 2014. In 55 European centres more than 1000 patients will be sequenced in the next year and a half.

If you have questions about MYO-SEQ or would like to participate, please contact Monica Ensini (monica.ensini@ncl.ac.uk)

The CTSR has proven here quite clearly its capability to galvanize interest and collaboration efforts that can then be translated into action in a fast and effective way.
THE GFB ONLUS WAS RECORDED IN THE REGIONAL REGISTER OF FAMILY SOLIDARITY ASSOCIATIONS

After a year of its establishment as a Volunteer Organization, the GFB ONLUS was also recorded in the regional register of Lombardy of Family Solidarity Associations.

ON THE SITE OF THE GFB ONLUS THE COUNT OF PATIENTS BELONGING TO THE GROUP

The GFB ONLUS has published on its website the count of patients so far registered in the group since 2010, with particular reference to the country of origin.
THE FIRST PHASE OF THE AMERICAN PROJECT ON THE LGMD2E HAS BEEN SUCCESSFULLY COMPLETED

In 2013 it has already been started, at the Research Institute by Nationwide Children’s Hospital - Columbus Ohio, the beta-sarcoglycan gene transfer project for treatment of Limb Girdle Muscular Dystrophy Type 2E.

Dr. J.R. Mendell and Dr. L. Rodino Klapac had started their work, the scientific way to be followed is based on their experiences, gained by completing a similar trial for the treatment, throughout a gene transfer therapy, of the Limb Girdle Muscular Dystrophy Type 2D. Dr. Mendell explained in details aims and phases of the pathway to be followed:

**Aim 1.** Determination of pre-clinical efficacy of the transfer of human b-sarcoglycan gene, using recombinant adeno-associated virus to act as delivery vehicle, in b-sarcoglycan deficient mice. Time required: one year.
**Aim 2.** Regulatory preparation for a “recombinant adeno-associated virus human b-sarcoglycan” gene transfer intramuscular clinical trial, including formal toxicology/biodistribution study and clinical vector production. Time required: about one year.

**Aim 3.** Perform an intramuscular clinical gene therapy trial with recombinant adeno-associated virus human b-sarcoglycan transfer (into the “extensor digitorum brevis” muscle) in LGMD2E patients. Time required: about one year starting since the closing of the previous aim.

*Today the first phase of the project has been successfully completed, in 2014 the second phase will be developed, so to reach the first clinical trial for beta-sarcoglycan in 2015.*

Moreover, Dr. Mendell underlined his clear intention to take this project to the following important level (Aim 4) which target is the vascular delivery of the missed gene to the lower limbs. He is in fact confident this can be achieved with an outcome that will mean widespread gene expression into the treated muscles and functions improvement. At this purpose they are already testing and discussing, with U.S. FDA, a procedure which will be developed and carried out during the next eighteen months.

We wish to underline this trial-study, as we know up to now, is today the most important worldwide scientific project entirely and solely committed to the research of a treatment for LGMD2E. It is therefore extremely important for us and represents a substantial step in order to get more attention to the disease that affects our children. We can say it is a reward for all the efforts we have made till now, independently on the results and the applications it will produce.
THE GFB ONLUS HAS BEEN ENROLLED AT THE NATIONAL CENTRE FOR RARE DISEASES

The GFB ONLUS has been enrolled in the associations of patients' groups, at the National Institute of Health - National Centre for Rare Diseases.

The list can be found at the link:

http://www.iss.it/site/cnmr/dina/asso/as01.asp

A REGISTRY FOR PATIENTS LGMD2C
The American Foundation "Kurt + Peter Foundation" collects in its patient registry the data on patients with LGMD2C (gamma-sarcoglycan deficiency). This registration is done online at the link provided below. It is in 3 parts and lasts about 15 minutes. The answers will be very useful for the analysis of variable progression of this disease and several other features specific to the LGMD2C. We recommend you to complete the form in its entirety, although some of the questions are optional.

http://www.kurtpeterfoundation.org/patient_registry


The GFB NGO will present a poster in electronic format at the European Congress of Rare Diseases ECRD2014, which will take place in Berlin, in the days 8-9-10 May, where the latest updates regarding the sarcoglycanopathies will be presented. From July 7 to 10, the researcher Paola Bonetti, a member of the Committee of the GFB medical and scientific non-profit organization, will present the poster of the association in Nice, at the International Congress of Neuromuscular Diseases ICNMD2014.
THE SECOND CLINICAL TRIAL OF GENE THERAPY FOR LGMD2D HAS BEEN STARTED IN COLUMBUS OHIO.

Prof. J. Mendell, at Nationwide Children's Hospital in Columbus Ohio USA, started in October the second clinical trial of gene therapy for the LGMD2D on six patients, which will end in March 2016. No patients will be recruit for this trial.

Great emotion and satisfaction for all our GFB members.
More info from:


THE LGMD2D FOUNDATION
News

The newly formed LGMD2D Foundation officially launched their website on October 8th, 2013. The website will host the patient registry, provide information about the disease and upcoming studies, discuss relevant scientific discoveries and provide researchers with the information necessary to apply for grants.

www.lgmd2d.org

THE PROJECT OF CROWDFUNDING IN SUPPORT OF GFB ONLUS
In the month of September it was started the PROJECT of Crowdfunding on the Platform Youcaring of the GFB ONLUS, to promote the scientific research on LGMD2E and to organize a Second International Conference on sarcoglycans and other dystrophies of the gridles.

All information about the project can be found at the following link. You can help GFB ONLUS throughout a donation by Paypal or by a credit card and by turning this message to all your contacts.

http://www.youcaring.com/nonprofits/nothing-is-impossible-help-us-to-treat-/89954

YOU CAN SUPPORT US BY ◾ BANK TRANSFER, PAYPAL, CREDIT CARD OR BY OUR T-SHIRTS

BANK TRANSFER
These are the bank accounts of the Family Group of Beta-sarcoglycanopathies non-profit organization (NPO GFB) on which to make donations to support the fight against the beta-sarcoglycanopathies and the other forms of muscular dystrophy:

Bank account: Poste Italiane

HEADING: Gruppo Familiari Beta-sarcoglicanopatie Onlus

IBAN IT41O0760111000001012232672 BIC / SWIFT code BPPIITRRXXX

Bank account: Credito Valtellinese

HEADING: Gruppo Familiari Beta-sarcoglicanopatie Onlus

IBAN IT71J052165227000000002059 BIC / SWIFT BPCVIT2S

DONATE BY PAYPAL OR CREDIT CARD
News

Read the instructions on www.beta-sarcoglicanopatie.it

T-SHIRT OF GFB ONLUS

Available sizes: 3-4, 5-6, 7-8, 9-10, 11-12, S, M, L, XL, XXL. Offer from 10 €. Home delivery free of charge. To order write to info@beta-sarcoglicanopatie.it

ON THE INTERNET ALL THE VIDEOS ON THE FIRST NATIONAL CONFERENCE GFB
All the videos of the First National Conference GFB have been published on the site www.lgmd2e.org

GFB NPO was born

On February 27, 2013 the GFB NPO (Beta-sarcoglycanopathies Family Group Onlus), was born.

It has been registered to the registration number SO-81 of the provincial section of Sondrio in the regional register of voluntary organizations in Lombardy, Italy.
President: **VOLA BEATRICE**  Vice-President: **PERLINI MARCO**
Fiscal code: **91015450140**

**Donations by bank**

This is the account of the Beta-sarcoglycanopathies Family Group Onlus (GFB NPO) on which you could make donations to support the fight against beta-sarcoglycanopathies and other forms of muscular dystrophy:

IBAN code: **IT41O0760111000001012232672**  
BIC / SWIFT code: **BPPIITRRXXX**

**Support us**

Any amount you want to give for the research will be a real help in the fight against beta-sarcoglycanopathies and other forms of muscular dystrophy.
On Thursday, May 23rd, the GFB ONLUS took part in the National Uildm Events in Lignano Sabbiadoro, in the parallel session:

"Meeting of the Beta-sarcoglycanopathies Family Group"

Conducted by: Beatrice Vola, President of the GFB Onlus

"The meeting is intended to give a space to the interested people, who can talk and discuss together, to introduce the GFB Group and its objectives, to plan our activities for the year 2013."

Time: 15-18 - Sala Rosa, Le Vele

ROBERTO MAGGI ELECTED MEMBER OF NATIONAL COUNCIL UILDM
On Saturday, May 25th, prof. Roberto Maggi, a member of the medical-scientific committee of GFB Onlus, was elected member of National Council UILDM for the period 2013-2015.

http://www.uildm.org/2013/05/26/eletta-la-nuova-direzione-nazionale-uildm/

April 19th: FIRST NATIONAL CONFERENCE GROUP FAMILY BETA - SARCOSGLYCANOPATHIES

SARCOCGLYCANOPATHIES: What next?

Sarcoglycanopathies and limb girdle muscular dystrophies, scientific research and clinical aspects

GFB is inviting you to the first national convention of Beta-sarcoglycanopathies Family Group where various issues related to the scientific research and clinical aspects of limb girdle
muscular dystrophies will be treated. It will be in Milan, on Friday, April 19th, 2013. Oral presentations will be mostly in Italian language but probably with also a simultaneous translation into English and the direct stream. The final program and the registration form will be in the next newsletter.

For more info:


Among the many speakers at the conference:

- **Jerry R. Mendell**, MD   Professor of Pediatrics and Neurology

  Director, Center for Gene Therapy

  Director, Paul D. Wellstone Cooperative Muscular Dystrophy Research Center

  The Research Institute at Nationwide Children's Hospital, Columbus Ohio USA

- **Louise Rodino-Klapac**, Center for Gene Therapy   Principal Investigator

  Neuromuscular Disorders   Principal Investigator
The Research Institute at Nationwide Children's Hospital, Columbus Ohio USA

Massimiliano Cerletti, Group Leader

Principal Investigator/Muscle Stem Cell Unit

Progenitor Labs Ltd (GlaxoSmithKline)

London BioScience Innovation Centre  London, United Kingdom

PhD Principal Investigator Harvard-GlaxoSmithKline Skeletal Muscle Program

Department of Stem Cell and Regenerative Biology

Harvard University and Harvard Stem Cell Institute Cambridge, MA USA

Robert Pleticha, Online Patient Communities Manager, EURORDIS (rareconnect)

Saverio Tedesco, Senior Research Associate

Cell & Developmental Biology, Div of Biosciences, Faculty of Life Sciences
University College London, London, United Kingdom

Anna Ambrosini, Research Program Manager

Telethon Foundation, Milan

Giacomo Comi, Department of pathophysiology and medical-surgical transplant, University of Milan

Doriana Sandonà, Faculty: Medicine and Surgery

Research group: ER processing of skeletal muscle membrane proteins, Department of Biomedical Sciences, University of Padua

Claudio Semplicini, Department of Neuroscience, University of Padua

Gian Maria Fimia Ph.D. Cell Biology Laboratory, National Institute for Infectious Diseases, Lazzaro Spallanzani IRCCS Rome

FROM NOVEMBER GFB IS A MEMBER OF “ALLIANCE TREAT-NMD”
THE O.Ma.R HAS PUBLISHED A PRESS OF GFB

The O.Ma.R (Italian Observatory rare diseases) on 29th November 2012 published a statement “Beta-sarcoglycanopathies, the Group of GFB families seeks other Italian patients with this disease.”

AT THE START THE GENE THERAPY PROJECT FOR LGMD2E

It has already been started, at the Research Institute by Nationwide Children’s Hospital - Columbus Ohio, the beta-sarcoglycan gene transfer project for treatment of Limb Girdle Muscular Dystrophy Type 2E, whose news we had already given you months ago.

Even before receiving the research grant necessary to finance the execution of the first phase, Dr. J.R. Mendell and Dr. L. Rodino Klapac had started their work following the pathway they had shown us. The scientific way to be followed is based on their experiences, gained by completing a similar trial for the treatment, throughout a gene transfer therapy, of the Limb Girdle Muscular Dystrophy Type 2D.

We wish to underline this trial-study, as we know up to now, is today the most important worldwide scientific project entirely and solely committed to the research of a treatment for LGMD2E. It is therefore extremely important for us and represents a substantial step in order to get more attention to the disease that affects our children. We can say it is a reward for all the efforts we have made till now, independently on the results and the applications it will produce.

Dr. Massimiliano Cerletti, member of our group’s scientific committee, is willing to closely follow the activities Dr. Mendell and his staff will carry out. In November Dr. Cerletti will be in Columbus to discuss this project.

If you or anybody else is interested in this project and wishes to get more information about it, please contact us to: info@beta-sarcoglycanopatie.it
CLINICAL STUDY AT THE CIVIL HOSPITAL IN PADUA ON LGMD2E

At the Civil Hospital of Padua, in these days is going to start a one-year clinical trial on beta-sarcoglycans, in co-operation with the research groups of the Institute of Myology in Paris and the University of Copenhagen. Professor Elena Pegoraro is the coordinator of the project in Italy, Dr. Pascal Laforet is the contact for France and Dr. John Vissing for Denmark.

The aim of the project is to study the clinical evolution of beta-sarcoglycans or LGMD2E in thirty patients, who for a year will make some visits to assess the cardiac, respiratory and muscular situation.
Some patients enrolled in the GFB are participating in this study. For more information please contact the telephone number 393280075986.
A gene therapy medicine with adeno-associated virus has been recommended for authorization in the European Union for the first time.

Glybera (alipogene tiparvovec), developed by uniQure, a Dutch biotech, is designed for patients with the genetic disorder lipoprotein lipase deficiency (LPLD) who have severe or multiple pancreatitis attacks, despite dietary fat restriction. The medicine is administered as a single injection.

MIRACLE FOR THREE

What would you do if someone told you that all three of your children have a rare and sometimes fatal genetic disorder (sarcoglycanopathy) and that they will be confined to wheelchairs for the rest of their lives? We are the Zubairs and we refuse to accept that life sentence for our children. We need you help. Support us in finding the best treatment for our children.
A STUDY ON LGMD2C at Genethon INSTITUTE OF PARIS

Important oral presentation on gamma-sarcoglycanopathies Congress ESGCT "European Society of Gene and Cell Teraphy", that was held in Versailles 26 to 29 October 2012 (page 46):

Monitoring by serum miRNA gene transfer of a treatment in a mouse model g-sarcoglycanopathy
David Israeli, Genethon, Evry

GFB HAS PARTICIPATED IN THE WORKSHOP "RARE DISEASE AND ORPHAN DRUG Registries"

The GFB has participated in the International Congress on the records of rare diseases and orphan drugs, organized by EPIRARE in Rome, on 8th and 9th October 2012.

http://www.epirare.eu/meet/20121008.html

POLL EPIRARE REGISTERS ON PATIENTS IS ON-LINE

It's online the poll Eurordis for all the patients who need to express their views and expectations on the records of rare diseases patients. There are still some weeks to fill it in. Please, anyone who has a rare disease fill it in as soon as possible.

GENE THERAPY FOR LGMD2E

Dr. Jerry R. Mendell confirms his availability to perform a gene replacement therapy clinical trial to restore a normal copy of the human β-sarcoglycan gene to the muscle of patients with LGMD2E.

We recently spoke, by phone conference-call, with Dr. Mendell, MD nearby the Nationwide Children’s Hospital, Columbus Ohio, U.S.A. He confirmed to us his interest and availability to duplicate researches and clinical trial already performed on gene therapy approach for the clinical treatment of LGMD2D, focusing this time the attentions on LGMD2E gene therapy.

During the conversation and afterwards with a document he wrote and delivered to us, Dr. Mendell explained in details aims and phases of the pathway to be followed:

**Aim 1.** Determination of pre-clinical efficacy of the transfer of human β-sarcoglycan gene, using recombinant adeno-associated virus to act as delivery vehicle, in β-sarcoglycan deficient mice. Time required: one year.

**Aim 2.** Regulatory preparation for a “recombinant adeno-associated virus human β-sarcoglycan” gene transfer intramuscular clinical trial, including formal toxicology/biodistribution study and clinical vector production. Time required: about one year.
**Aim 3.** Perform an intramuscular clinical gene therapy trial with recombinant adeno-associated virus human b-sarcoglycan transfer (into the “extensor digitorum brevis” muscle) in LGMD2E patients. Time required: about one year starting since the closing of the previous aim.

Moreover, Dr. Mendell underlined his clear intention to take this project to the following important level (Aim 4) which target is the vascular delivery of the missed gene to the lower limbs. He is in fact confident this can be achieved with an outcome that will mean widespread gene expression into the treated muscles and functions improvement. At this purpose they are already testing and discussing, with U.S. FDA, a procedure which will be developed and carried out during the next eighteen months.

If anybody is interested with this project and wish to know more about it, please take contact with us writing to: info@beta-sacrcoglicanopatie.it

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**EXPLORATORY PROJECTS FOR GENETICS DISEASE MOST NEGLECTED**

From Telethon new funds on research of neglected disease even among those rare: in fact, have been funded eleven projects that focus on genetic disease.

(read more)

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**New study of pluripotent stem**
Based on the IPSC, the induced pluripotent stem cells, the new work of the team led by Francesco Saverio Tedesco and Giulio Cossu, Department of Cell and Developmental Biology at University College London, lays the groundwork for a new strategy for muscular dystrophies.


ECRD Congress - Brussels

On Wednesday, May 23 2012, the GFB was in Brussels at the sixth European Conference of Rare Disease. On Thursday the GFB has presented the poster number 92, entitled "Beta-sarcoglycanopathy: any longer an orphan disease?"
Dr. Thomas Voit discusses Gamma Sarcoglycan Gene Therapy :: March 2012

Dr. Thomas Voit, Professor of Pediatrics at the University of Pierre and Marie Curie, Paris 6, and Scientific and Medical Director of the Institute of Myology in Paris, discussed Gamma Sarcoglycan Gene Therapy, about a phase I trial of adeno-associated virus serotype 1-γ-sarcoglycan gene therapy for limb girdle muscular dystrophy type 2C.

Brain.

APPEAL

JOIN US: in order to support our goals it’s really important to give the widest diffusion of this bulletin that follows, through your e-mail contacts, facebook and messanger.

“To the supporters of the struggle against the beta-sarcoglycanopathy

We are a group of family members of people with beta-sarcoglycanopathy, which is a rare form of Limb girdle muscular dystrophy and we formed this Italian group a few months ago.

You can help us giving the widest diffusion of this press, by sending it to your e-mail contacts or facebook’s friends.
In recent years we realized there aren’t precise scientific studies, aimed at therapeutic approaches targeted for this disease. ......."

RECENT STUDY ON THE MURINO MODEL

An important research on beta-sarcoglycan mice was coordinated in Italy by Dr. Maraldi N.M. at the University of Bologna and was funded by the Ministry of Education, Universities and research in 2008.

Title of the research is “Molecular mechanism in muscular dystrophy with cardiomyopathy and potential therapeutic approaches”. The study has been divided into 5 research areas. The beta-sarcoglycanopathy were treated by the chief scientist M. Sampalessi at the University of Pavia and by L. De Angelis at the University “La Sapienza” of Rome.