

## **GFB □ □ BETA-SARCOGLYCANOPATHY FAMILY GROUP**

**ABOUT US:** We are a group of families with people with beta-sarcoglycanopathy and other Limb-girdle muscular dystrophies, rare forms of muscular dystrophy. Our group started with some families in Lombardy. During the years it was so enlarged that now other people living in the whole Italy and abroad have joined us.

### **OUR OBJECTIVES:**

THE GFB ONLUS was created to finance research projects aimed at treating the disease.

It wants to be a way of keeping us/you better informed on info/news not easy to be found as well as the most updated specific research activities for this particular disease. Our mission is **to represent people with Beta-sarcoglycanopathy**, towards the institutions, research organizations, other associations, to patients.

This role is kept only by us till now, because no other groups exist with our same purpose - neither in Italy nor abroad.

This website and its self-mutual-help group GFB ONLUS were created from the desire to get in

touch with other families affected by this disease (on the Italian territory they are about fifty), so to share the various aspects of living every day with this disease, as well as clinical, legal, logistical, organizational aspects related to scientific research and the use of various aids.

Our intention is to realize a first collection of data on these patients, in view of a future patient register and specific clinical trials.

**DISEASE AND RESEARCH:** In the years we have experienced the disease of our children, realizing there were no precise scientific studies and therapeutic approaches targeted for this disease (until 2012). We were completely left alone. The disease, as defined by the European Union and the European Association for rare diseases (E-RARE), was seen as “neglected” for the almost total absence of both research and diagnostics on European territory.

There is now a model of beta-sarcoglycan mouse produced in the United States and only one publication - [pubblicazione](#) - of the research on murine model. Our group GFB ONLUS (beta-sarcoglycanopathies family group ONLUS) has been running since 2011 to promote the scientific research on this specific pathology.

Since the month of August 2012 the families of GFB have decided to start the first fundings to the [project of gene therapy for LGMD2E](#) Prof. J. Mendell in Columbus Ohio. **Five payments for a total of \$ 1,300,000** have been paid up today

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In these five years the GFB has received five reports with all the results of the [phases of the project](#) and has taken part in seven conference calls with American doctors and the medical and scientific committee of the association.

**REGISTRATION:** On February 27<sup>th</sup>, 2013 the GFB Onlus was officially created in Italy as a voluntary organization , registered to the provincial section of the regional register of voluntary organizations in Lombardy n. SO-81.

In 2014 it was also included in the regional register of the Associations of Family Solidarity n. 947.

**Referents of the Group:** Vola Beatrice and Perlini Marco  
Chiara Galloni (secretary) 0039 3493374060  
Email volbeat@libero.it, info@beta-sarcoglicanopatie.it  
Headquarters: Italy

## [ARTICLES AND PUBLICATIONS](#)



## **PATIENTS RECORDED IN THE GFB ONLUS**

## PATIENTS WITH LGMD2E

provincia di residenza

o stato

n pazienti

2010

n pazienti

2013

n pazienti

2014

n. pazienti

2015

n. pazienti

2016

n. pazienti

2017

MILANO ITALIA

1

1

1

LECCO ITALIA

2

2

2

SONDRIO ITALIA	3	3	4
TREVISO ITALIA	2	2	2
TRENTO ITALIA	1	1	1
ANCONA ITALIA	1	1	1
ROMA ITALIA	1	1	
BRESCIA ITALIA	1		
STATI UNITI	1	1	3
IRAN	1	1	1
GERMANIA	1	1	1
INDIA	1	1	1
GRECIA	1	1	1
OLANDA	4	4	4
PORTOGALLO	1	1	1

SPAGNA	2	2	2
CANADA	2	1	1
PALESTINA	1	2	
LIBIA	1	1	
REGNO UNITO	1	1	
TURCHIA	1	3	
SVEZIA	1	1	
FRANCIA	1	1	
SUD AFRICA	1		
DANIMARCA	1		
ARGENTINA	1		
COLUMBIA	3		
BRASILE	1		
<b>TOTALE</b>	<b>5</b>	<b>14</b>	<b>21</b>

**PATIENT WITH LGMD2D**

o stato

n pazienti

2010

n pazienti

2013

n pazienti

2014

n. pazienti



2015

n. pazienti

2017

TREVISO	1	1	1
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GENOVA	1	1	1
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BRINDISI	1	1	2
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REGGIO CALABRIA	1	1	1
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MODENA	1	1
MILANO	1	1
CAGLIARI	1	
ROMA	1	
ALESSANDRIA	1	

BRASILE	1	1	2
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BELGIO	1	1	1
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OLANDA	3	12	15
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GIAPPONE	1	1	1
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FINLANDIA	1	1	2
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INGHILTERRA	1	2	8
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GERMANIA	3	3	3
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SPAGNA	2	8
UNGHERIA	1	1
STATI UNITI	5	22
CANADA	1	1
PAKISTAN	3	3
ARGENTINA		

1			
COLOMBIA	1		
AUSTRIA	1		
FRANCIA	2		
IRAN	4		
COSTARICA	1		
AUSTRALIA	2		
GIORDANIA	1		
RUSSIA	1		
ALTRO	3	5	9

<b>TOTALE</b>	1	15	28
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**PATIENTS WITH LGMD2C**

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provincia di residenza

o stato

n pazienti

2010

n pazienti

2013

n pazienti

2014

n. pazienti

2015

n. pazienti

2017

VICENZA	1	1	1	1
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BRESCIA	1	1	1	1
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PALERMO	1	1	1
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PADOVA	1	1
TORINO	1	1
REGGIO EMILIA	1	
LATINA	1	
PESCARA	1	
TOSCANA	1	

STATI UNITI	2	2	2
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SERBIA	1	1	2
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CANADA	1	2	2
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AUSTRALIA	1	1	1
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FRANCIA	1
FILIPPINE	1
TUNISIA	2
ALGERIA	1
TURCHIA	1
GERMANIA	1
IRAN	8

ALTRO

1

1

6

TOTALE

4

9

12

37

### PATIENTS WITH LGMD2F

provincia di residenza

o stato

n pazienti

2010

n pazienti

2013

n pazienti

2014

n. pazienti

2015

n. pazienti

2017

INDIA

1 1 1 1

IRAN

3

**TOTALE**

1

1

1

4

## PATIENTS WITH INDEFINITE SARCOGLYCANOPATHY

provincia di residenza

o stato

n pazienti

2010

n pazienti

2013

n pazienti

2014

n. pazienti

2015

n. pazienti

2017

CALTANISSETTA ITALIA

3

**TOTALE**

3



**Together we can**