**BIO-NMD: searching for biomarkers to improve the development of treatments and diagnosis of people affected by neuromuscular disease**

What is BIO-NMD?

BIO-NMD is an EU funded research project focusing on:
- Duchenne muscular dystrophy;
- Becker muscular dystrophy;
- Ullrich congenital muscular dystrophy;
- Bethlem myopathy

The project is searching for biomarkers in people with these conditions – an explanation of biomarkers is given in the box below.

What are Biomarkers?

Biomarkers are substances in the body that offer a way to measure normal or abnormal processes. This means that processes associated with particular diseases can be measured and disease progression monitored. It also means that the effect of drugs or other therapies on disease progression can be evaluated.

A biomarker may be a protein found in bodily fluids such as the blood or urine, or in tissues such as the muscles. Alternatively, a particular gene could be a biomarker - measuring the activity of certain genes may be able to give an accurate picture of disease progression.

Why have these conditions been chosen for the study?

Neuromuscular disorders include a large number of different conditions so studying them all at once would be impractical and expensive. The BIO-NMD researchers therefore chose Duchenne and Becker muscular dystrophy which are both caused by changes in the dystrophin gene and Ullrich congenital muscular dystrophy and Bethlem myopathy which are both caused by changes to collagen VI genes as a starting point. They chose these conditions because the genetic defects causing them are well characterised, and there is already a body of information available on how these genetic defects cause the symptoms.

These conditions are also representative of many others because they include early and late onset forms, fast and slow progression and severe and mild symptoms. In addition, they affect two different parts of muscle cells which are also often affected in other conditions, so any biomarkers found may be more widely applicable.

By focusing on a small number of conditions, efforts can be intensified to find biomarkers which can then be applied to other conditions. This was true for example during research into biomarkers for breast cancer which have then proved useful for other cancer types.

Who is involved in BIO-NMD?

There are 12 European partners involved in the BIO-NMD project. Some are well placed to collect and distribute patient samples and others are experts in studying genes or proteins, data analysis, animal models or project management.

The project is coordinated by Prof. Alessandra Ferlini at the University of Ferrara in Italy. The other partners are: Leiden University Medical Centre; University of Newcastle upon Tyne; University of Padova; Inst. of Child Health, University College London; University of Rome Tor Vergata; INSERM Montpellier; University of Milan; Royal Institute of Technology, Stockholm; Ariadne Genomics Inc.; Applied Biosystems; Novamen.

Who can tell me more?

BIO-NMD is working with representatives from the patient community in the form of a Patient Association Committee. This committee can be contacted through the BIO-NMD website at www.bio-nmd.eu.

Alternatively, the project’s communication officer, Cathy Turner at Newcastle University, can be reached on catherine.turner@ncl.ac.uk

BIO-NMD’s Patient Association Committee:
- Marita Pohlschmidt, Muscular Dystrophy Campaign UK
- Elizabeth Vroom, United Parent Project Muscular Dystrophy
- Anna Ambrosini, Telethon Italy

How might this benefit people living with neuromuscular disease?

A major application of new biomarkers is in clinical trials. At the moment, when a new drug is being tested, researchers use a variety of ways to measure whether the drug has had a positive effect. However, these measures are not always very good at showing small changes and improvements in symptoms, especially when the drugs are only being tested for a short time.

However, if the BIO-NMD project can find biomarkers in patients’ blood or urine, samples of these can be taken throughout clinical trials. Measuring the levels of these biomarkers will show researchers clearly and accurately whether the drug being tested has had an effect or not.

Other benefits of biomarkers include:
- Blood and urine testing may be able to replace the use of muscle biopsies in the future.
- Diagnosis can happen earlier because testing for biomarkers is quicker and easier than genetic testing.
- Disease progression can be accurately measured allowing better clinical management of symptoms.
- Existing treatments can be adjusted to precisely meet the needs of individual patients and ensure they get the maximum benefit.