



# Royal College of Physicians

## Professor Francesco Muntoni Biography

Francesco Muntoni graduated in Italy in 1984 and completed his training in Child Neurology and Psychiatry in 1989. Since his degree he worked on childhood neuromuscular disorders, initially in Italy and, since 1993, in London, where he worked at Imperial College/ Hammersmith Hospital until 2007. At Imperial College he directed the Neuromuscular Centre following the retirement of Professor Victor Dubowitz in 1996. In 2008 he moved to the UCL Great Ormond Street Institute of Child Health in London with his entire clinical, academic and diagnostic groups. His areas of interest include the clinical, molecular genetic and biochemical aspects of childhood neuromuscular diseases.

Original contribution to research include the discovery that the X-linked dilated cardiomyopathy (XLDCM) is secondary to a dystrophin gene abnormality (allelic mutations in dystrophin cause Duchenne muscular dystrophy); the identification of more than 30 novel disease genes, ranging from congenital myopathies, to congenital dystrophies to limb girdle muscular dystrophies.

More recent areas of research relate to the regulation of splicing control of genes involved in neuromuscular disorders. In particular Muntoni is involved in research both in the control of splicing of the SMN gene involved in spinal muscular atrophy; and in antisense oligonucleotide induced exon skipping in Duchenne muscular dystrophy, and in allele specific silencing of dominantly inherited neuromuscular genes. The more advanced program of research is the one on the therapeutic trials regarding the administration of antisense oligonucleotides in Duchenne muscular dystrophy. After having completed a series of clinical trials aimed at developing the antisense oligonucleotide for skipping exon 51, the consortium led by Muntoni is currently performing a multicentre trial aimed at skipping exon 53 with a novel antisense oligonucleotide in Duchenne muscular dystrophy, funded by the European Community and Sarepta Therapeutics. Muntoni also leads a UK multicentre translational research network focused on trial readiness for SMA in UK, with the capturing of disease progression and validation of novel outcome measures.