



Duchenne
UK

Research Portfolio End of Year Report 2024



JoiningJack.



Thank you Joining Jack!

Thank you for your continued support and invaluable contributions to date towards our efforts to end Duchenne muscular dystrophy (DMD).

The purpose of this report is to provide an end-of-year progress update on all active and completed projects funded by Joining Jack in the Duchenne UK portfolio. Please note that projects completed in 2023 and before are not included.

This report will also share our focus for 2025 which is always informed by patient unmet needs and gaps in Research & Development that we feel we are uniquely placed to address.



Joining Jack 2024 highlights



Joining Jack is supporting us in our efforts to target the Duchenne heart and accelerate access to new treatments with **£200,000** committed to our Therapeutic Grant Call and **£100,000** to our Help the Heart Grant Call.



Joining Jack is contributing to 10 active projects in the DUK portfolio (including cardiac projects).

- 1 project is progressing towards newly agreed timelines.
- 3 projects will start in the New Year.
- 4 projects have been completed.



Our financial reconciliation is being completed. We will be in touch should funds require reallocation.



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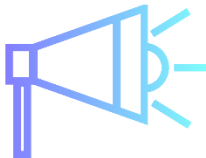
We are pleased to share that in 2024 Duchenne UK have also...



Committed to 13 projects across medical research, care, and access to treatments



Received 56 new research enquiries, engaging us with new companies



Kicked off the Duchenne Patient Data Platform Project starting with the relaunch of the In Case of Emergency (ICE) app!

‘I feel very proud of what we have achieved in 2024, with your invaluable support. We are committed to identify and fund the best projects that can transform DMD treatment and care, to achieve better outcome for all people living with Duchenne.’



Dr Alessandra Gaeta
Director of Research and Development, Duchenne UK

Active Projects

DUK 2020-08 DMD Hub Manager

Institution: University of Newcastle
Principal investigator: Emma Heslop
Status: Ongoing until December 2025
Funding contribution: £ 5,000

This funding is supporting Emma Heslop's post as Project Manager of the DMDhub. Emma is central to developing and expanding the remit of the DMD hub whilst supporting the existing established initiatives. A 12 month no cost extension was granted this year to continue the hub's aims.

We are pleased to share that since 2016 the DMD hub has:

- Engaged with 16+ companies and 5 Clinical Research Organisations
- Recruited 793 boys on DMD trials
- Facilitated 57 clinical trials. 28 are currently active, 7 at feasibility stage and 4 in early discussions
- Provided 34 posts to facilitate the day to day and set up of new trials. 72% of these posts have been sustained
- Leveraged £1.3 million of funding from other funders and industry to support DMD clinical research

DUK 2021-02 Hydrotherapy

Institution: Lancashire Teaching Hospital
Principal investigator: Christian De Goede, Consultant Paediatric neurologist
Status: Ongoing until December 2025
Funding contribution: £118,000

This project aims to evidence the benefit of hydrotherapy treatment in DMD through a clinical trial. Hydrotherapy is a form of exercise that involves doing exercises with a physiotherapist in a heated swimming pool.

3 trial sites are involved, including:

Manchester (fully recruited), Preston (fully recruited) and Oswestry (recruiting).

Based on the current recruitment rate, the team are on track to reach the recruitment target of 44 across 3 sites early this year. We are excited to look at the data, which if positive will provide evidence of the value of this intervention and a case for it to be provided to boys with DMD.

DUK 2021-07 Innate immune response

Institution: Binghamton University, New York
Principal investigator: Nagaraju Kanneboyina

Status: Awaiting final report
Funding contribution: £1,000

This study looks at the body's innate immune responses and stopping it from mounting a response to AAV Gene Therapies, which is a major barrier to re-dose these therapies in DMD and prevents some boys from accessing these treatments, if they have pre-existing antibodies to AAV.

A no-cost extension request was granted last year to allow for the completion of experiments proposed in the project aims whilst publications were under review. The aims were taking longer than expected to complete, due to the complex nature of the animal models used but we are due to receive a final report by the end of the year which will draw final conclusions.

DUK 2022-01 PhD – Antifibrotic screening platform and testing

Institution: University of Newcastle
Principal investigator: Jordi Diaz-Manera, Professor of Neuromuscular, Translational Medicine and Genetics
Status: Ongoing until August 2025
Funding contribution: £32,688

Funding supports a 3-year PhD post which has already made substantial progress.

Fibroadipogenic precursor cells (FAPs) are the main cells responsible for the expansion of fibrotic and fatty tissue in muscles of patients with Duchenne muscular dystrophy (DMD). FAPs can convert into cells that are responsible for fibrosis in DMD, called fibroblasts and adipocytes, and therapies that are able to stop this process could slow disease progression. This laboratory has experience in the isolation and characterization of FAPs from healthy people and DMD patients.

The team have completed the first aim in the project of characterising the potential toxic effect of several selected drugs in vitro and are now completing trials in DMD mouse models to test their potential effects as antifibrotic drugs.

DUK-2022-02 PhD – Neuropsychiatrist post/psychosocial WS

Institution: Devon Partnership Trust
Principal investigator: Rory Conn, Consultant child & adolescent Psychiatrist

Status: Ongoing until July 2025

Funding contribution: £75,000

The intention for the 1st year of this post was to induct Rory into the DMD network and upskill him on the condition and evidence base to date for psychiatric involvement which was successful.

Rory has now completed his second year, which has involved the dissemination of his clinical advice and support, assessment (both online and in person) of the young people who fall under the project aim and developing the basis of the clinical guidelines, in collaboration with the DMD Care UK psychosocial working group, which are currently at draft stage and expected to be finalized soon.

Rory's monthly online surgeries for clinicians to come together to discuss cases and themes relating to psychosocial care have been successful and have provided key insights. Typically, there are around 5 attendees per surgery, bring cases and themes, as wide ranging as educational dilemmas to sex and sexuality.

DUK 2022-10 SMART SUIT (Elevex)

Institution: Duchenne UK

Status: Ongoing

Funding contribution: £432,051

In 2024, the team identified the need for stronger internal control and a deeper integration of user perspectives, so brought the Elevex design work in-house. We now have a dynamic, collaborative team, comprising engineers, biomechanics experts, and control systems specialists. Crucially, young people from the DMD and SMA communities have joined the design team and have actively shaped the latest concept development work, building on learning from the previous prototypes.

December 2024 saw us achieve a significant milestone with the unveiling of an alpha prototype of the groundbreaking under-arm Elevex concept, which has already received encouraging initial feedback from stakeholders.

Early 2025 will focus on gathering detailed feedback from clinicians and the DMD community.

This invaluable input will inform the next phase: refining the mechanical architecture and developing an intuitive control system for effortless user operation.

DUK 2023-01 Developing nutritional guidance, resources and a structured nutritional programme

Institution: University of Glasgow

Principal investigator: Jarod Wong, Consultant Paediatric Endocrinologist

Status: Ongoing until January 2027

Funding contribution: £204,073

The aim of this project is to develop guidance and materials to support better diet and nutritional management in boys with DMD and to gather evidence, through research, of the impact of steroids on weight.

Progress in each part of the project has successfully started to:

- Identify the timing of weight gain in boys with DMD following initiation of steroids – collection of retrospective data is expected to be completed by 2025.
- Identify the caloric needs of both ambulant and non-ambulant boys on steroid therapy – The NutrInD study is now recruiting in Glasgow.
- To identify the opinion of young people with DMD, their carers and health care providers in the UK – the team is analysing data from an online survey to inform the workstream of the nutrition working group of DMD Care UK, which is closely involved with this project.

The results of the above ongoing research studies as part of this funding will provide the key information and guidance for the development of standards of care and educational resources on nutrition. The first completed draft of the clinical recommendations of the DMD care UK nutrition working group is expected in the summer of 2025.

DUK 2023-04 DMDhome validation

Institution: Aparito

Principal Investigator: Elin Haf Davies

Status: Ongoing until September 2026

Funding contribution: £168,336; £5,000 relocated to care December 2023

The aim of this project is to validate DMD Home, a new video assessment of motor function, including ambulation and arm function, as a tool that companies could use to assess the effect of their treatments in DMD clinical trials. We are particularly interested in validating this to assess 1) the transfer stage, which is the stage of transitioning from the ambulatory to the non-ambulatory stage, when boys are still able to bear weight while transferring, and 2) arm function. Being able to assess these would enable non-ambulant boys to access more clinical trials, as there would be a way to measure the impact of new drugs in this population.

The study involves filming short videos from the comfort of your home every 6 months for 24 months. This is a novel approach using video

capture and computer vision analysis to measure both arm and leg function.

Recruitment for this study is currently open.

DUK 2024-03 DMD Care UK Phase 2

Institution: University of Newcastle

Principal investigator: Michela Guglieri

Status: Ongoing until June 2026

Funding contribution: £169,988.53

Your funding is supporting DMD Care UK to continue to deliver its ambitious programme of work to drive the development, dissemination and adoption in the NHS of expert guidance for all areas of DMD clinical management including psychological support and consolidating its impact to date to achieve real-world benefits for people living with DMD.

Building on its success since its launch in 2019, in its second phase, the programme will also deliver more resources that families will be able to access for free to help them feel more empowered and supported in their journey with DMD.

DUK 2024-09 DMD Care UK Transition Project

Institution: Leeds Teaching hospital trust

Principal investigator: Anne-Marie Childs

Status: Ongoing until September 2027

Total FFF funding contribution: £200,000

Entering adulthood is a key stage in anyone's life but can be more complicated when you have DMD. The failure to support young people with complex health needs as they move into adulthood has been highlighted in a number of high-profile publications.

The team are developing a robust young-adults DMD transition care pathway that addresses the clinical and psychosocial needs of this group, as well as identifying ways to improve transition in different settings. This should encourage services providers to recognise and use this DMD Care model, as we will include appropriate 'cost neutral' suggestions for service improvements in different settings.

Completed projects

DUK 2018-27 Neuromuscular Complex Centre

Institution: University of Newcastle

Principal investigator: Chiara Marini-Bettolo

Status: Completed

Funding contribution: £10,000

The ultimate aim is to establish a Neuromuscular Complex Care Day Unit for multidisciplinary management of neuromuscular patients from North East and Cumbria in England where Standards of Care appointments will be scheduled over a 1-2 day admission.

A survey (PDSA cycle 1) circulated nationally to patients with neuromuscular diseases and their caregivers showed that overall patients are supportive of such a model of care and allowed us to better shape this initiative to meet their needs.

- 107 patients and care givers took part in the survey
- Patients on average attend 4.7 specialist appointments a year
- 19% of patients have had an unplanned hospital admission in the last 12 months
- 45.8% of patients report that their physiotherapy needs are not met in the community
- On average 37.4 % of patients travel 30-60 minutes to attend their clinic appointment
- 69.8% of patients and 48.1 % of parent/care givers prefer to have their neuromuscular care appointments scheduled over one day
- 80% of patients would be keen to attend such a unit, should this be available to them

DUK 2022-08 Leeds Research Fellow

Institution: Leeds Teaching Hospital

Principal investigator: Anne-Marie Childs

Status: Completed

Funding contribution: £35,000; £7,362.47 reallocated to care December 2023

The research fellow supported the day-to-day running of research trials and assisted the creation of new ones; two studies have been initiated and a further 3 are expected to open within the first 3 months of 2025!

The research fellow had a key role in raising awareness about DMD research with paediatric trainee doctors and across the wider multi-disciplinary teams, ensuring that each and every child could be

considered for enrolment in a research study and no opportunities were missed.

DUK 2023-02 DMD Care UK project manager

Institution: University of Newcastle
Principal investigator: Michela Guglieri
Status: Completed
Funding contribution: £22,245

DMD Care UK has now successfully opened working groups (WG) across all agreed SoC (Standards of Care) areas now 13 in total.

Each WG is working towards building consensus on their area of SoC through drafting within the WG, consultation across all NS centres (with neuromuscular consultants as well as relevant specialists in that area of care). Several WGs have completed this stage and have published their guideline, endorsed by the relevant professional body (cardiac, respiratory, bone and endocrine).

Patient and family guides to the SoC have been produced in parallel with publication of each clinical guideline and in consultation with the Family Focus Group of DMD Care UK. This enables patients/families to better advocate for their care and to bring latest information to the attention of their clinical teams as well as to make informed choices about their care.

DUK 2024-01 DMD Care UK Physio WG Consultancy

Institution: Anna Mayhew Consultancy
Status: Completed
Funding contribution: £4,000

The overarching aim of the project was to improve the delivery of therapy to those living with DMD in the UK.

Anna has successfully completed the following:

- Re-writing and disseminating the survey as the results we had were pre-pandemic
- Published a paper on 'Reporting on delivery of standards of care for Duchenne muscular dystrophy in UK and guidelines for improving implementation'
- Created and published associated therapy guide on delivering DMD SoC in UK
Created and published associated parental guide on delivering DMD SoC in UK

The recommended next steps for this project included the dissemination to the community through DMD Care UK and other platforms/routes which we are progressing.



Our focus for 2025

Continuing to fund research to help the Duchenne heart



Cardiac complications, including sudden heart failure, are the primary causes of mortality in DMD, sometimes in very young patients.

We want to stop this.

Driving technology innovation



Ensuring people with DMD have the products to give them the freedom and confidence to move through life and embrace whatever comes their way!

Building Duchenne UK's Data platform



Robust, complete and high-quality longitudinal DMD patient reported data is lacking. Our goal is for patients to have the data and insights to empower them to make informed decisions, giving them the confidence and freedom to navigate their care journey. We will integrate our flagship programmes (DMD Care UK and DMD Hub) to unlock their full impact.



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Thank you!

We are so grateful to Joining Jack and all your supporters for your relentless drive in our shared fight to end Duchenne. We hope that this report highlights the incredible impact and progress that your funding has enabled. Duchenne UK is enormously proud to have Partner Charities like you. Together, we will end Duchenne.

For any queries related to this document, please contact research@duchenneuk.org



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Duchenne UK, Unit G24, Shepherd's Building, Charecroft Way, Hammersmith, W14 0EE duchenneuk.org Charity number: 1147094