

(digital PCR) OR (dPCR)

NCT Number		Title	Authors	Description	Identifier	Dates
1	pubmed:35897138	Dystrophinopathy Phenotypes and Modifying Factors in DMD Exon 45-55 Deletion	Javier Poyatos-García Pilar Martí Alessandro Liquori Nuria Muelas Inmaculada Pitarch Luis Martinez-Dolz Benjamin Rodríguez Lidia Gonzalez-Quereda Maria Damiá Elena Aller Marta Selva-Gimenez Roger Vilchez Jordi Diaz-Manera Jorge Alonso-Pérez José Eulalio Barcena Amaia Jauregui Josep Gámez Jesus Angel Aladrén Ariadna Fernández Marisol Montolio Inmaculada Azorin David Hervas Ana Casasús Marisa Nieto Pia Gallano Teresa Sevilla Juan Jesus Vilchez	OBJECTIVE: Duchenne muscular dystrophy (DMD) exon 45-55 deletion (del45-55) has been postulated as a model that could treat up to 60% of DMD patients, but the associated clinical variability and complications require clarification. We aimed to understand the phenotypes and potential modifying factors of this dystrophinopathy subset.	pmid:35897138 doi:10.1002/ana.26461	Thu, 28 Jul 2022 06:00:00 -0400
2	pubmed:36067829	Prevalence and Detection of Actionable BRAF V600 and NRAS Q61 Mutations in Malignant Peripheral Nerve Sheath Tumor by Droplet Digital PCR	Erica Y Kao Kristina M Wakeman Yu Wu John M Gross Eleanor Y Chen Robert W Ricciotti Yajuan J Liu Jose G Mantilla	Malignant peripheral nerve sheath tumors (MPNSTs) are aggressive tumors with poor prognosis that do not typically respond well to standard chemotherapy. Recently, point mutations involving BRAF V600E have been demonstrated in a subset of MPNST, offering the possibility of targeted treatment. However, the reported prevalence of these alterations is variable. Mutations involving NRAS, which is also involved in the MAPK/ERK pathway and amenable to targeted inhibitors, have not been well...	pmid:36067829 doi:10.1016/j.humpath.2022.08.005	Tue, 06 Sep 2022 06:00:00 -0400