Results This study reports the result of the validation process. In total there were 132 participants. Content validity was achieved by the nominal group technique. In this study, technology enhanced quality of life questionnaire (EITVAQ) is a feasible tool, having a high satisfaction rate of 78%, a response rate of 76.5% and an average completion time of 4 mins and 26 seconds. EITVAQ had a score of 0.92 (Cronbach Alpha). When comparing with the control, the two lowest scores were the social and emotional aspect. The two major differences in scores among both groups were the physical and cognitive well-being. All participants who participated in the TELP managed to complete the task learning the skills of creating a music video.

Conclusion EITVAQ, an interactive and child-friendly tool to assess quality of life is now validated. It aims to be used widely among children with hydrocephalus, providing a baseline assessment to allow us to understand more about a child’s quality of life from their own perspective. This study concludes that technology has a huge potential in helping children with hydrocephalus and various neuro-disability to integrate into society.

GP125 MY LIFE, MY VOICE: TECHNOLOGY-ENHANCED QUALITY OF LIFE ASSESSMENT TOOL FOR CHILDREN WITH HYDROCEPHALUS
Joy Tan, John Caird, Alf Nicholson. Temple Street Children University Hospital, Dublin, Ireland; RCSI, Dublin, Ireland; Beaumont Hospital, Dublin, Ireland

Background To date, children with hydrocephalus continue to have a considerable long-term outcome. However, current literature on health-related quality of life (HRQOL) among children with hydrocephalus are limited. This serves a call for research to validate a suitable HRQOL for children with hydrocephalus measuring the physical, emotional, social and cognitive well being.

Objective To validate a technology-enhanced quality of life questionnaire (EITVAQ) as an effective assessment tool measuring the current well-being of a child with hydrocephalus (child-centred).

Methods This is a prospective study which took place since January 2018 to January 2019. This study received ethical approval during the current well-being of a child with hydrocephalus.

Participants and methods The study included 63 boys, aged from 6 months to 8 years with elevated creatinine phosphokinase (CPK), according to laboratory tests. After medical genetic counseling molecular genetic analysis was performed for all patients. The MLPA method was used to search for deletions and duplications in the DMD gene, the analysis of point mutations was carried out by NGS, if the MLPA method did not reveal pathogenic variants. Sanger sequencing was used to validate mutations identified by the NGS.

Results Totally, in 39 patients we revealed different alterations in DMD gene. Among them 11 (28%) had a point mutation. It was 4 nonsense, 4 missense, 2 splicing mutation and one single-nucleotide duplication. Five mutation were novel. They are splicing c.10798 – 2A > G, missense c.22887 > A (p.Val763Asp) and c.3269A > T (p.Gln1090Leu), nonsense c.858T > G (p.Tyr286X) and duplication c.8325dup (p.Glu2776Thrfs*6). The remaining 28 (72%) patients had gross duplications 3 (8%) and gross deletions 25 (64%) in the DMD gene. Interestingly, more than half of the patients had deletions in the region of exons 45–51 of the DMD gene.

Conclusion Our study showed that the most common cause of Duchenne dystrophy in Russian children are gross deletions of the DMD gene, in particular deletions in the region of exons 45–51 occurring most frequently.