Study ID 202502_TE

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Date of Approval: March 2025
Name of study: Growth, General development and Gonadal function and their relationship to Genetics and rhGH therapy in Noonan Syndrome (NS-5G)
Study period: Mar 2025 – Mar 2028
Summary Recombinant human growth hormone (rhGH) has been shown to improve growth in NS. However, the growth response to rhGH may vary depending on the genotype. Furthermore, NS can affect several systems including cardiac, metabolic and tumour development. Current evidence does not indicate that rhGH therapy adversely affects the risk of any unwanted effects in NS. However, more long-term studies are needed in sufficiently large cohorts and the current study aims to use GloBE-Reg to perform a long-term observational study of the safety and efficacy of rhGH in children with NS and link it to the genetic variations that are found in cases of NS.
Inclusion criteria – All patients with genetically confirmed or suspected NS at all existing and new centres using the GloBE-Reg Registry.
Exclusion criteria – No.
Data to be collected for all participating cases: Core Data Core Data History GloBE-Reg rhGH Minimum Dataset (retrospective and/or prospective) - Diagnosis Case Report Form (CRF) - Therapy CRF – One visit per 6-month period while on daily or long-acting rhGH therapy. For participants not receiving or yet to start rhGH, data should still be collected once every 6-months. - Clinician Reported Outcome CRF – One visit per 6-month period while on rhGH therapy and, where available, prior to rhGH initiation.
Data cut timelines: 6 Monthly data tranches with deadlines in March and September

Expected outputs These will be disseminated by a publication in a journal, presentation in scientific meetings or in GloBE-Reg newsletter.
Publication Plan for authorship in outputs The investigator will adhere to the GloBE-Reg recommendations. All current and future participating centres will be co-authors on all planned publications as long as they meet the ICMJE criteria listed. The coauthors will include those involved in study design, data provision, data extraction, data analysis and interpretation and those involved in the creation of the outputs. It is possible that as the study group matures, research into specific outcomes may be driven by different study group members. The support of all funders of the GloBE-Reg platform at the time the study was active will be acknowledged.