

European Medical Education Initiative on Noonan Syndrome

Background and planned activities

The **European Medical Education Initiative on Noonan Syndrome** (Noonan syndrome and related disorders) aims to raise awareness of this rare disorder and ultimately improve patient care by assessing disease management across Europe, identifying gaps in current clinical practice and suggesting means with which to optimise diagnostic, therapeutic and monitoring procedures. This project is an independent initiative driven by a Steering Committee comprised of experts from different specialties across Europe (full list provided at the end of the document), with support by an unrestricted grant from Novo Nordisk Europe A/S. Novo Nordisk will have no influence on any of the scientific content or material that will be generated as part of this initiative. The educational grant will be used for the purposes of logistical and medical writing support by a Medical Communications Agency, as well as for compensation of the Steering Committee members for participation in remote working meetings in relation to the development of the survey (see below).

In order to obtain a “snapshot” of current clinical practice, the members of the Steering Committee are in the process of developing a **clinical practice survey** focusing on the diagnosis and clinical management of diseases within the Noonan Syndrome phenotypic spectrum. This survey will be distributed throughout Europe, with the aim to obtain feedback from relevant patient management centres across the continent, focusing on geneticists, paediatric endocrinologists and paediatric cardiologists. The Steering Committee will assess the survey results, with the ultimate goal of developing a publication for submission to a peer-reviewed scientific journal. The questionnaire is currently in development and the survey is expected to be available by the end of July. The deadline for completing the survey online will be end of September 2020.

To ensure a comprehensive response to the clinical survey, the Steering Committee is seeking the support of pan-European and National specialist societies for human genetics, paediatric cardiology and paediatric endocrinology. To this end, we would like to discuss with you the feasibility of cooperating in the distribution of the survey. If so, **we would appreciate the Society’s response in terms of possible ways to distribute the survey**, such as:

- (a) The Society distributes the link to the survey directly to its members in a separate correspondence
- (b) The Society distributes the link to the survey to its members as part of a newsletter/general correspondence
- (c) The Society provides a list of key contacts that the Steering Committee can approach

Given the timelines involved, a prompt response would be greatly appreciated.

Steering Committee members and supporting agency

Chair

- Thomas Edouard, Paediatric Endocrinologist, Toulouse, France

Steering Committee members

- Emma Burkitt-Wright, Clinical Geneticist, Manchester, United Kingdom
- Sixto García-Miñaur, Geneticist, Madrid, Spain
- Jan Lebl, Paediatric Endocrinologist, Prague, The Czech Republic
- Ingegerd Östman-Smith, Paediatric Cardiologist, Gothenburg, Sweden
- Guftar Sheikh, Paediatric Endocrinologist, Glasgow, United Kingdom
- Marco Tartaglia, Geneticist, Rome, Italy
- Alain Verloes, Geneticist, Paris, France

European Noonan Syndrome Medical Education Initiative

Briefing document

June 2020

- Cordula Wolf, Paediatric Cardiologist, Munich, Germany
- Martin Zenker, Geneticist, Magdeburg, Germany

Supporting medical communications agency

- Physicians World Europe GmbH, Mannheim, Germany