NEUROFIBROMATOSIS

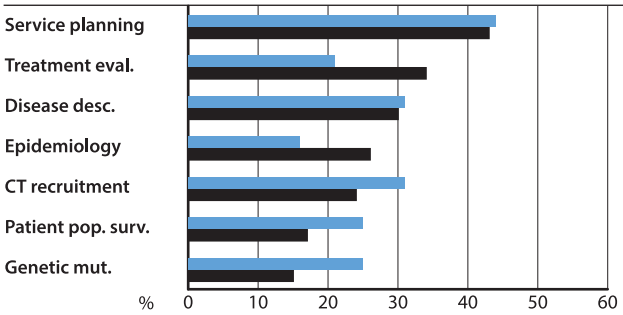
CLINICAL PICTURE

Neurofibromatosis (NF) is a genetic neurological disorder that can affect the brain, spinal cord, nerves and skin. Tumours, or neurofibromas, grow along the body's nerves or on or underneath the skin. There are three types of neurofibromatosis. Neurofibromatosis type 1 (NF1) causes skin changes and deformed bones and usually starts at birth. Neurofibromatosis type 2 (NF2) causes hearing loss, ringing in the ears and poor balance. It often starts in the teen years. Schwannomatosis is the third and rarest type and less well understood in terms of natural history and inheritance (mostly sporadic). Schwannomatosis causes intense pain. Usually the tumours are benign, but sometimes they can become cancerous. It is the rarest type and it is.
There is no cure for NF and treatment is aimed at controlling symptoms. Depending on the type of disease and how bad it is, treatment may include surgery to remove tumours, radiation therapy and medicines. Type 1, in particular, is so varied in its manifestation, that it is difficult to predict outcome, as phenotype is so variable even within affected families. Most people with NF1 lead relatively long and healthy lives, but it does reduce life expectancy by around 15 years.
he major complications are hypertension and malignancy. NF2 generally has a worse prognosis. Much of the morbidity from these tumours results from their treatment.
Early detection and prompt attention to complications may reduce overall morbidity and mortality.

Age of onset	
Prevalence	+
Genetic Nature (inheritance)	
Number of Regional, National, European or International Registries	25

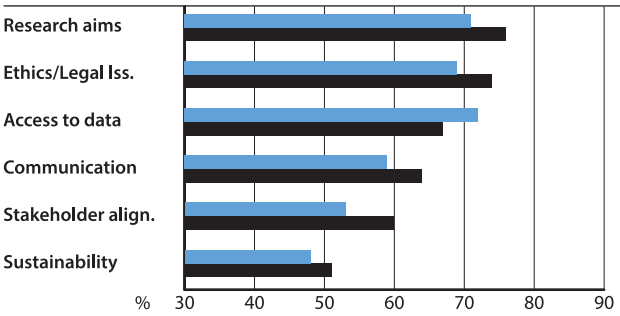
PARTICIPANTS IN THE SURVEY

The countries most represented amongst respondents concerned with NF include Denmark (19), Greece (12) and Italy (6).
A total of 54 survey participants reported being concerned with NF. The results below highlight major differences in opinion from NF respondents compared to other disease groups. As such, only a few results specific to NF are presented. For the remainder of survey questions, NF respondents did not differ significantly in their responses.



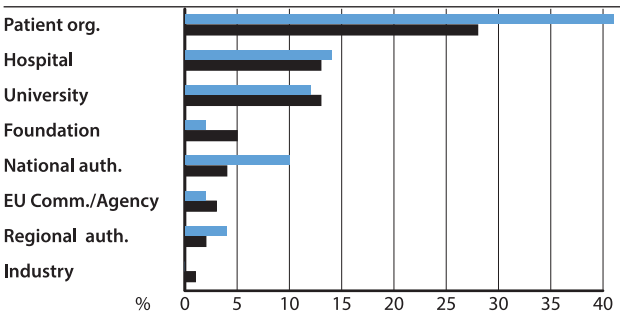
AIMS OF A REGISTRY

NF respondents less frequently reported a preference for treatment evaluation (20.8%) and more frequently for genetic mutations (24.5%)



REGISTRY GOVERNANCE

Overall, NF respondents less frequently reported the importance of a patient perspective in the governance of a registry as compared to other respondents except for the aspect of access to data (71.9%)



INITIATIVE FOR ESTABLISHING A REGISTRY

NF respondents that knew of the existence of a registry for their disease, more frequently reported patient organisations (40.8%) and national authorities (10.2%) as initiators of a registry as compared to other respondents.