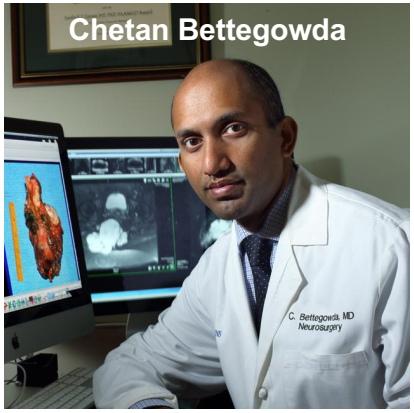
REiNS Disease Biomarkers Working Group

Winter 2017 Meeting

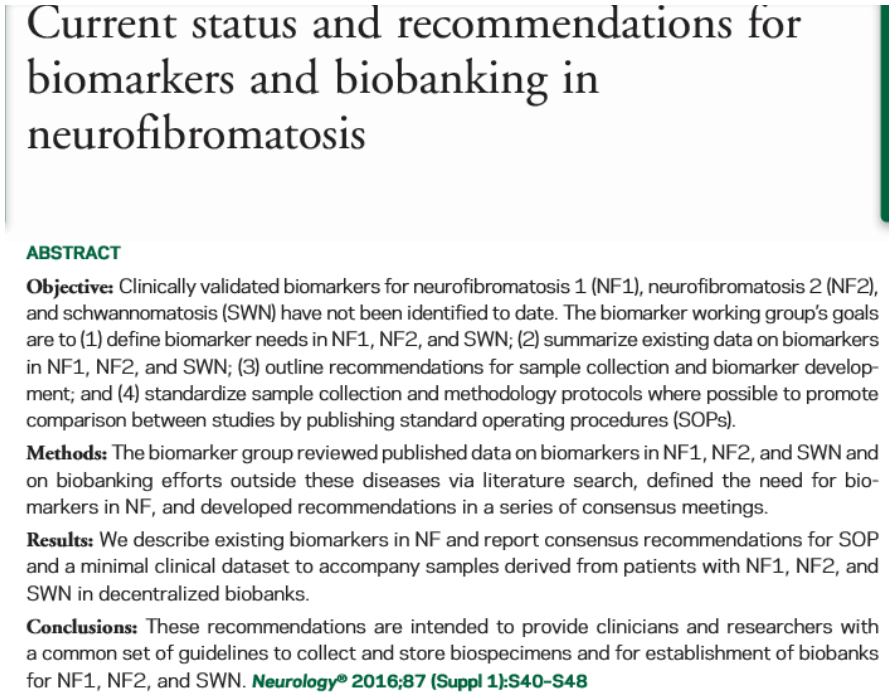
December 4, 2017



Response Evaluation In Neurofibromatosis Schwannomatosis
INTERNATIONAL COLLABORATION

Goals of the biomarkers group (2014)

- To define biomarker needs and summarize existing data in NF1, NF2, and Schwannomatosis
- To outline recommendations for sample collection and biomarker development
- To standardize sample collection and methodology protocols where possible to promote comparison between studies by publishing standard operating procedures



Current status and recommendations for biomarkers and biobanking in neurofibromatosis

ABSTRACT

Objective: Clinically validated biomarkers for neurofibromatosis 1 (NF1), neurofibromatosis 2 (NF2), and schwannomatosis (SWN) have not been identified to date. The biomarker working group's goals are to (1) define biomarker needs in NF1, NF2, and SWN; (2) summarize existing data on biomarkers in NF1, NF2, and SWN; (3) outline recommendations for sample collection and biomarker development; and (4) standardize sample collection and methodology protocols where possible to promote comparison between studies by publishing standard operating procedures (SOPs).

Methods: The biomarker group reviewed published data on biomarkers in NF1, NF2, and SWN and on biobanking efforts outside these diseases via literature search, defined the need for biomarkers in NF, and developed recommendations in a series of consensus meetings.

Results: We describe existing biomarkers in NF and report consensus recommendations for SOP and a minimal clinical dataset to accompany samples derived from patients with NF1, NF2, and SWN in decentralized biobanks.

Conclusions: These recommendations are intended to provide clinicians and researchers with a common set of guidelines to collect and store biospecimens and for establishment of biobanks for NF1, NF2, and SWN. *Neurology*® 2016;87 (Suppl 1):S40–S48

Genomic Biomarkers in NF Working Group

- Task
  - Describe genomic approaches to biomarker development in NF
  - Identify major research questions related to genomic biomarkers in NF
- Working Group
  - Co-Chairs – Oliver Hanemann, Chetan Bettegowda
  - Members
    - Jim Gusella (MGH/HMS)
    - Shawn Levy (HudsonAlpha)
    - Ludwine Messiaen (UAB)

Examples of genomic biomarkers

Genomic Approaches	Potential Application
Germline	
NF mutation	Genotype-phenotype correlations
Genetic Modifiers	Prediction of risk
Somatic	
Genomic sequencing	Indicators of progression/treatment targets
RNA sequencing	Indicators of progression/treatment targets
miRNA	Indicators of progression/treatment targets
Epigenomics	Indicators of progression/treatment targets
Liquid Biopsy	Early diagnosis of malignancy

Goals for 2018

- Identify and select two or three of the most pressing biomarker needs in NF
- Develop teams to help execute development of biomarkers
- Design experimental strategies to address these needs
- Commence sample collection, scientific experiments once appropriate ethics approval has been achieved

Patient Representatives

- Krizelle Alcantara
- Onno Faber
- Tracy Galloway
- Maquel “Mickie” Montgomery
- Herb Sarnoff