THERAPEUTIC AREAS OF INTEREST
RARE DISEASE | MEDICAL AFFAIRS

- *Gaucher Disease*
  - New diagnostic strategies
    - Electronic Health Records and Big Data analytics for early diagnosis
  - Clinical outcomes improvement for Cerezyme with focus on bone disease
    - Prognostic factors, treatment initiation, timing from diagnosis, biomarker trend, dose, adherence to treatment
  - Advance disease knowledge of Gaucher Disease type 3 (GD3)
    - Burden of disease, health economics and outcomes research in GD3
    - Advancing the understanding and management in GD3

- *Pompe Disease*
  - Pathophysiology of Pompe disease
    - Evolving phenotype of Pompe disease
    - Novel biomarkers in Pompe disease
    - Role of CNS
  - Impact of early diagnosis
    - NBS long-term follow up
    - AI enabled diagnostics
  - Effects of ERT
    - Long-term outcomes
    - Prognostic factors
    - Effect of transition between ERTs
  - Optimizing outcomes in Pompe disease
    - Optimal management of Pompe disease
    - Immune tolerance induction and management of patients with antibodies

- *Fabry Disease*
  - Pathophysiology of Fabry disease
    - Evolving phenotype of Fabry disease including sex related differences
    - Novel biomarkers & imaging assessments in Fabry disease
    - Genotype phenotype correlations
    - Treatment initiation decisions & role of pain
  - Impact of early diagnosis
    - Strategy for NBS
    - Implementation & utility of NGS panels
    - AI enabled diagnostics
  - Effects of Fabry specific therapy
    - Long-term outcomes
    - Prognostic factors
    - Effect of transition between therapies

sanofi

- *Acid sphingomyelinase deficiency (ASMD)*
  - Epidemiology of ASMD restricted to defining:
    - New High-Risk Population(s)
    - Birth prevalence through Newborn screening
  - Clinical approaches restricted to:
    - Development of biomarkers
    - Severity assessment tools and other prognostic tools
    - Olipudase alfa effectiveness
    - Disease progression in untreated patients
  - Pathophysiology of ASMD in humans

- *Cross Lysosomal Storage Diseases*
  - Diagnostic procedures for LSD: includes methods, algorithms, specific assessments
  - Tools to improve understanding of pathophysiology, disease severity and treatment outcomes
  - Newborn Screening for LSD
  - Epidemiology, prevalence studies in LSD
  - New therapeutic approaches for LSD: novel and combination therapies
  - Lysosomal cell biology and research studies
  - Safety and Efficacy of Immune Modulation Therapies for LSD

- *Immune-mediated thrombotic thrombocytopenic purpura (iTTP)*
  - Permanent or long-term sequelae: including assessment of therapeutic or diagnostic interventions that can help mitigate those sequelae
  - Efficacy and safety of caplacizumab as a first-line therapy in conjunction with IST without TPE

- *Immune thrombocytopenia (ITP)*
  - Biomarkers of immune dysregulation in ITP/hematology and in association to response to rilzabrutinib
  - Beyond low platelet burden in ITP and its association with immune dysregulation (BTK pathway) and rilzabrutinib multi-immune modulation MOA
  - Combination therapy with rilzabrutinib in ITP
  - Explore rilzabrutinib’s effect in other indications in hematology

- *Alpha-1 Antitrypsin Deficiency (AATD)*
  - High risk targeted testing of COPD/emphysema populations for AATD, including family testing
  - Definition of characteristics that identify high-risk populations for AATD
  - Epidemiology of AATD
  - Identification of disease characteristics predicting rapid decline in AATD
  - Long-term survival outcomes in AATD
  - Identification of meaningful change in clinical outcome measures for AATD
  - Correlation between biomarkers and surrogate outcomes with mortality in AATD

- *Hemophilia A*
  - Preclinical and basic science studies
    - Novel hemophilia outcome measures including biomarkers and tools to assess sub-clinical bleeding, bleed resolution, inflammation, bone health
  - Clinical Research
    - Clinical management and outcomes with ALTUVIIIO in hemophilia A subpopulations (e.g., mild/moderate hemophilia, women and girls with hemophilia, aging people with hemophilia, physically active patients)
    - Biomarkers and novel assessments of disease progression including sub-clinical bleeding, bleed resolution, inflammation, and joint and bone health
    - Studies of immune tolerance and immune tolerance induction
  - Real world qualitative and quantitative studies
    - Assessment of treatment of ALTUVIIIO on patient and caregiver reported outcomes, such as psychosocial function, pain, and mental health
    - Studies of the impact of ALTUVIIIO on resource utilization in hemophilia A

- *Hemophilia A and B*
  - RWE on fitusiran effectiveness on QoL/physical activity and joint and bone health (MRI, U/S, Dexa Scan)
  - Real-world performance/accuracy of commercially available IVD assays used during fitusiran treatment
  - Inhibitor development and Immune tolerance induction (ITI) on fitusiran prophylaxis
  - Studies to understand the switch between fitusiran and other non-factor therapies
  - Studies of fitusiran in other RBD (vWD-3 and platelets disorders) including females (hemophilia and non-hemophilia)