Guidelines for type 1 Gaucher disease

1. Diagnosis
    a. The whole phenotypic spectrum in all age groups
    b. Differential diagnosis
    c. Criteria for diagnosis – clinical, laboratory
    d. Newborn screening
    e. Genetic counselling
    f. Disease awareness

2. Treatment and monitoring
    a. Criteria for starting treatment
    b. Standard of care in all age group
    c. Dose and dose escalation versus agreed outcome
    d. outcome measurement instrument
    e. Hemoglobin, platelets, visceral volumes
    f. Biomarker responses
    g. Home therapy

3. Outcome that are important to patients
    a) Quality of Life, PROMs tools
    b) Nutrition, growth
    c) Supportive care
    d) Transition to adulthood
    e) Psychological aspects

4. Diagnosis and management of comorbidities/disease related complications/life events
    a. Parkinson
    b. Malignancies
    c. Osteoporosis/Avascular necrosis
    d. Splenectomy guidelines
    e. Pregnancies
    f. Menopause

5. Universal issues related to Gaucher disease
    a) Universal access to treatment/treatment options
    b) Variability among countries
    c) Universal applicability
    d) Telemedicine
    e) extended support/satellites etc