

# The French LYSODIAG platforms for DBS kit

## DESCRIPTION: WHAT?

- The French DBS kits aims to overcome the diagnostic delay in rare metabolic diseases, with Local French Laboratories. This means to develop a partnership with local centers in order to:
- to help and facilitate diagnosis by providing a simple tool to use in routine care
- to deliver fast results for clinicians



Centogene Service was available since 2013 in France

## RATIONALE: WHY?

In order to help the diagnosis in Lysosomal Storage Diseases (LSD), Shire global team has set up a partnership with Centogene, one of the world's leading companies in the field of rare genetic disease diagnostics, based in Germany. The partnership is providing a simple DBS kits and very fast results to clinicians to help the LSD diagnosis in routine care.

In France, the Centogene DBS kits have been used since 2013, but in 2016, CETL\* sent a letter to Shire France GM to request the forbidding of carrying biological samples outside France.

The medical team focused on a local partnership in order to continue to support LSD patient diagnosis in France.

## ROLES AND RESPONSIBILITIES: WHO?

The French medical affairs team coordinate several advisory boards in order to explore how to pursue the screening activity with national biology network instead of Centogene.

The global patient diagnostic team was also involved in the AdBoard meetings.

## FRENCH LAB PARTNER(S): WHO?

**Two local labs are involved:**

**CHU Rouen with Pr Soumeya Bekri & CHU Lyon with Pr Roseline Froissart**

## ROADMAP: WHEN ?



3 meetings with French biologists(x7)

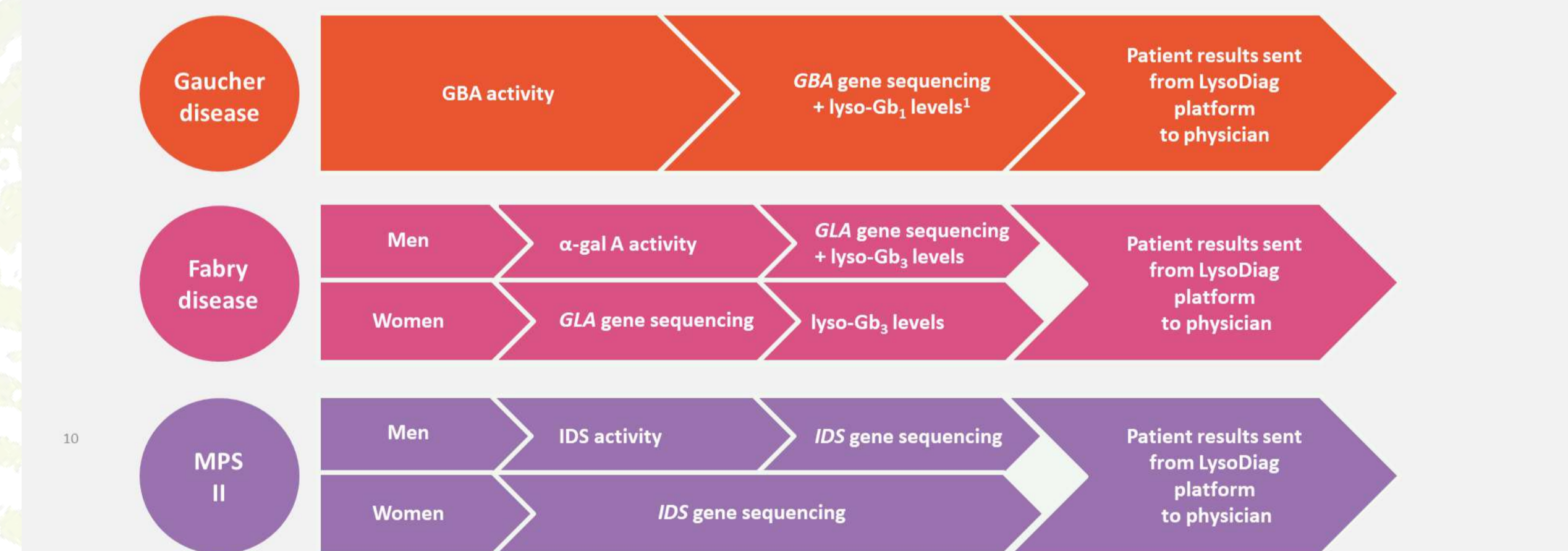
Accreditation of local laboratoires

Local and Global approval

Contrat negotiation

## TESTING PERFORMED

Lysodiag platform tests for Gaucher disease, Fabry disease and MPS I, II, III, VI & VII with the following analyses:



**Results:** This project will provide a local solution with an improved diagnostic support service to improve and enhance diagnosis in LSD in France and close the gap at National level.

\* (CETL: Comité d'Evaluation du Traitement des maladies Lysosomales - <http://www.cetl.net/>).