Patient Name: ARUPTEST, BCG GBA 1

Date of Birth: 05-Jul-15 Gender: Female ARUP ID: 10005

Requisition #: Client Supplied ID:

> Physician: ARUP, ARUP Printed: 23-Jun-17 09:24:47

ARUP Example Report Only 004848 500 Chipeta Way Salt Lake City UT 84108

Reported/ Procedure Result Units Ref Interval 17-174-900026 23-Jun-17 23-Jun-17 23-Jun-17 Gaucher GBA Activity Leukocytes Interp See Note f 08:46:00 08:46:00 09:22:59 Gaucher GBA Activity Leukocytes <0.2 L nmol/h/mg [4.6-12.0]17-174-900026 23-Jun-17 23-Jun-17 23-Jun-17 08:46:00 08:46:00 09:22:59

## 23-Jun-17 08:46:00 Gaucher GBA Activity Leukocytes Interp:

In this sample, beta-glucocerbrosidase (GBA) activity was undetectable, and consistent with a diagnosis of Gaucher disease (OMIM#230800, 230900, 231000). This test cannot predict disease severity. Additionally, this test does NOT evaluate enzyme activities deficient in other lysosomal storage disorders. Consideration should be given to DNA testing to confirm this finding. Genetic evaluation is recommended.

## 23-Jun-17 08:46:00 Gaucher GBA Activity Leukocytes Interp:

INTERPRETIVE INFORMATION: Gaucher Disease (GBA), Enzyme

Activity in Leukocytes

Gaucher Disease (GBA) Enzyme Activity in Leukocytes is reported in nanomoles hydrolyzed/hour/mg of protein.

Chart ID: 12272164