

SUPPLEMENTARY DATA

Effect of readthrough treatment in fibroblasts of patients affected with lysosomal diseases caused by premature termination codons.

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SUPPLEMENTARY FIGURE LEGENDS:

Supplementary Figure 1. Preliminary studies, testing gentamicin and PTC124 concentration, incubation time and monitoring cell viability in fibroblasts derived from P1 (Niemann-Pick AB), P5 (Sanfilippo B), P6 (Sanfilippo B) and a healthy individual. Monitoring of the residual enzymatic activities after gentamicin treatment during 24h, 48h and 72h (A) and PTC124 during 16h, 24h (B), 48h and 72h (C). Monitoring of the cell viability of P6 and a healthy individual after gentamicin (D) and PTC124 (E,F).

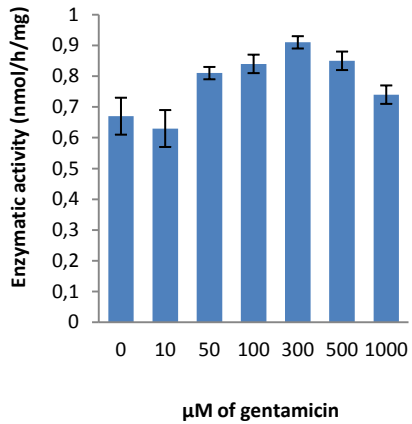
NPAB: Niemann-Pick AB disease, SF: Sanfilippo B disease.

A

24 hours

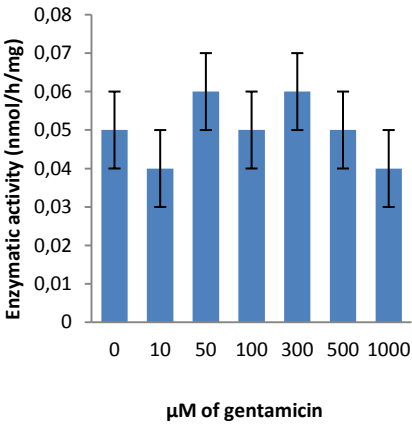
Patient 1

(P1), NPAB_24 hours



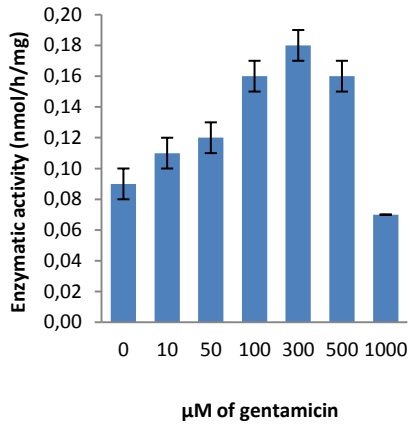
Patient 5

(P5), SF_24 hours



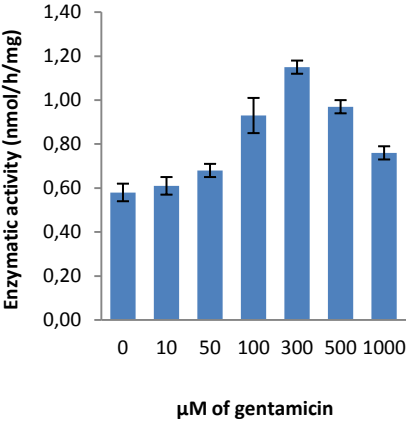
Patient 6

(P6), SF_24 hours

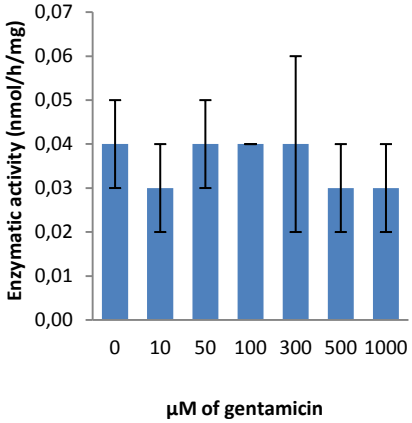


48 hours

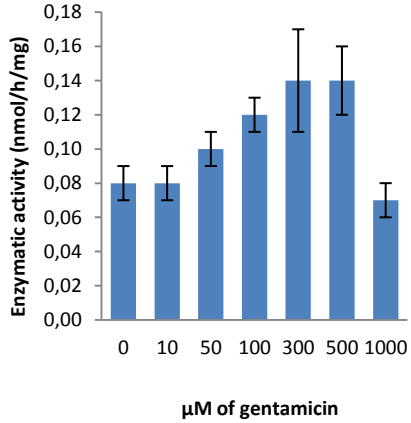
(P1), NPAB_48 hours



(P5), SF_48 hours

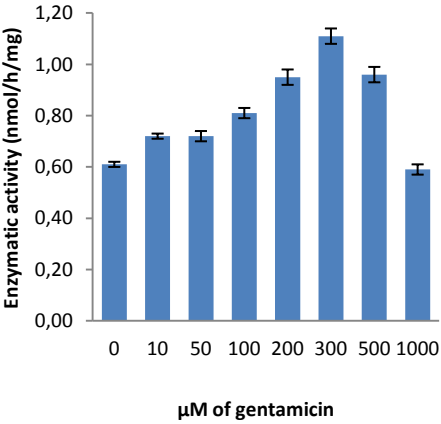


(P6), SF_48 hours

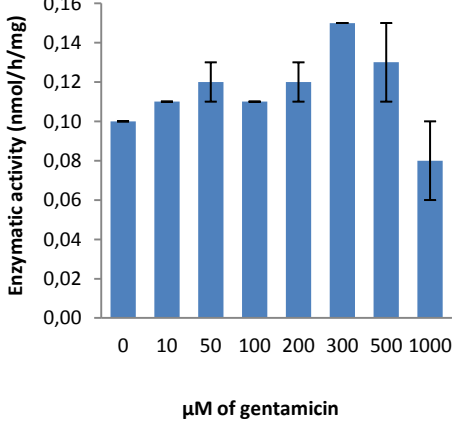


72 hours

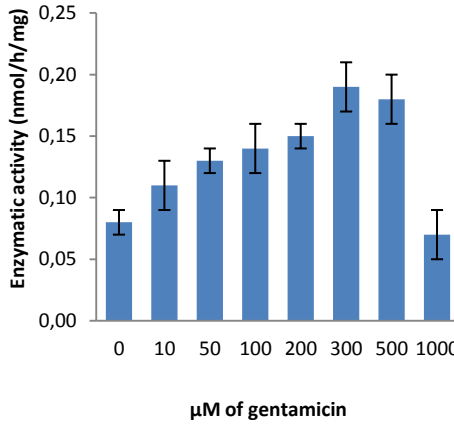
(P1), NPAB_72 hours



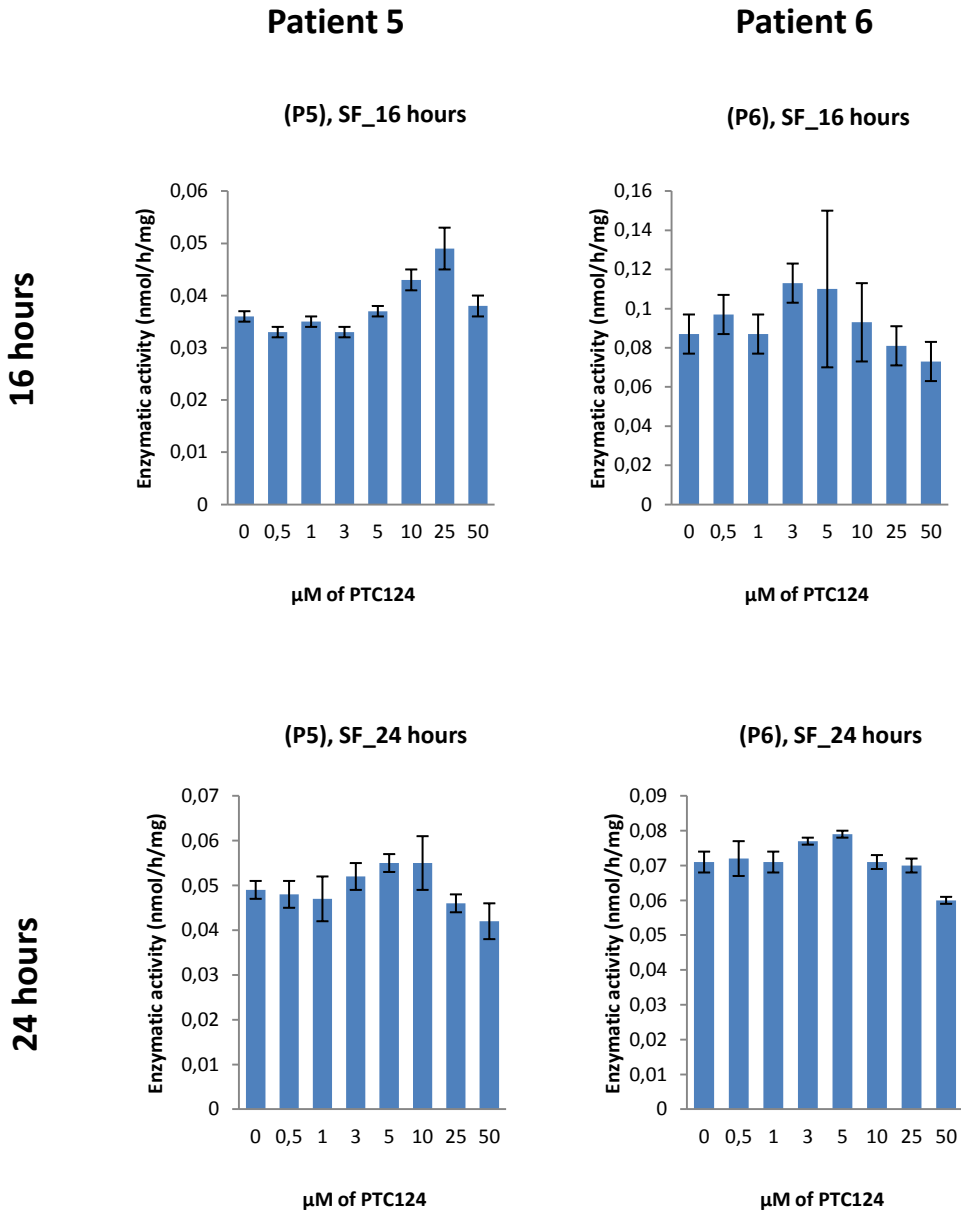
(P5), SF_72 hours



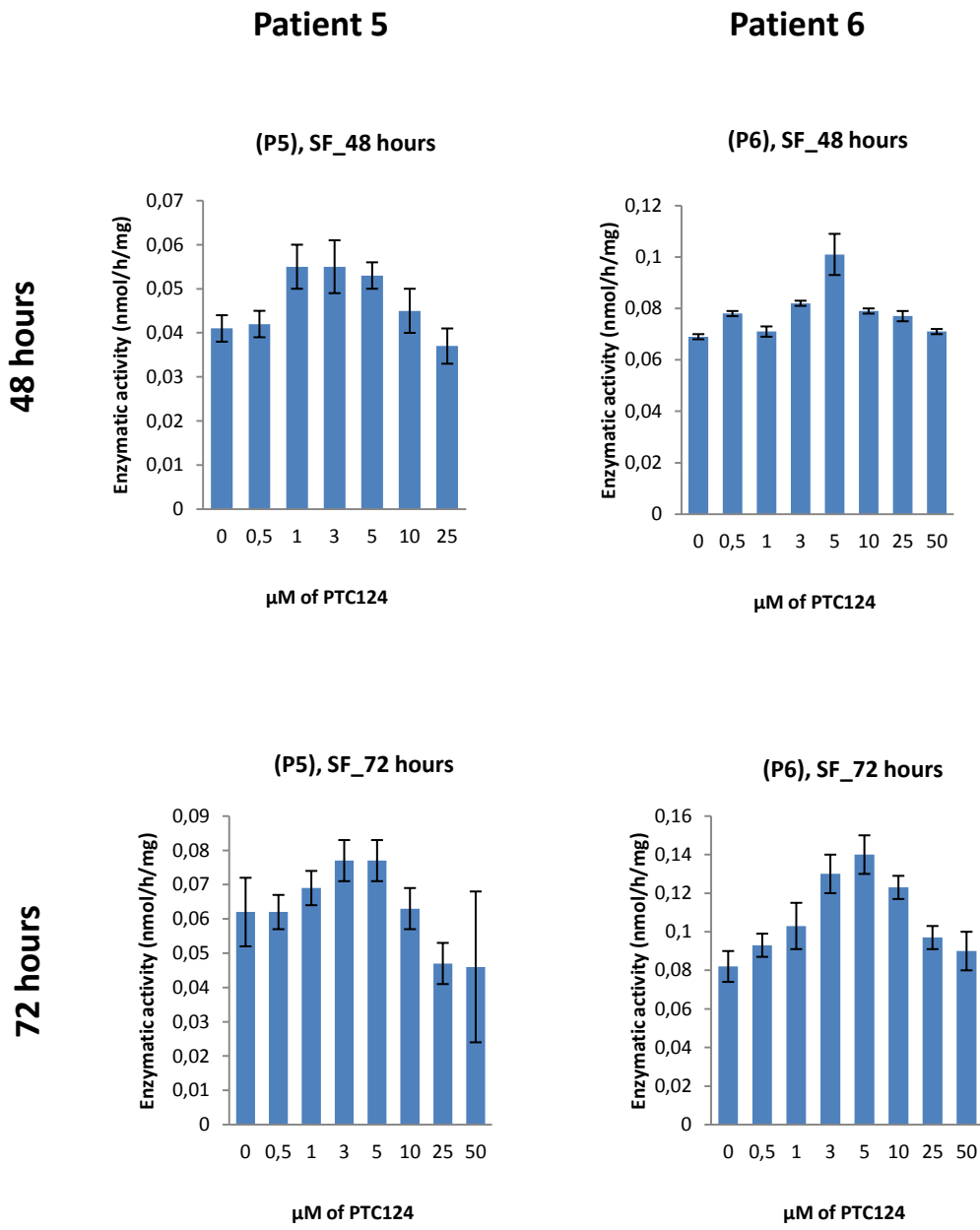
(P6), SF_72 hours



B



C

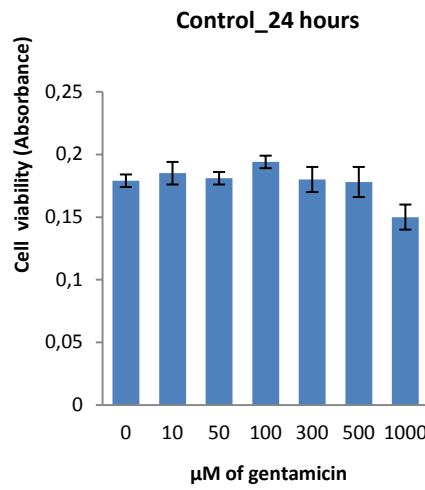
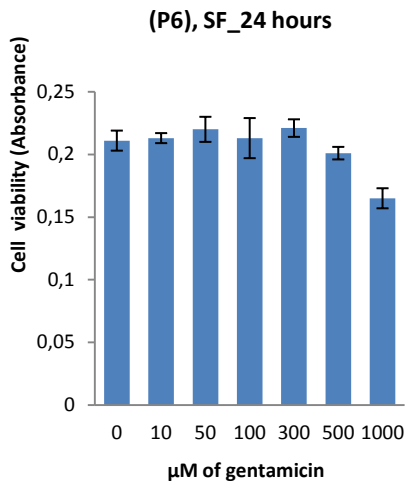


D

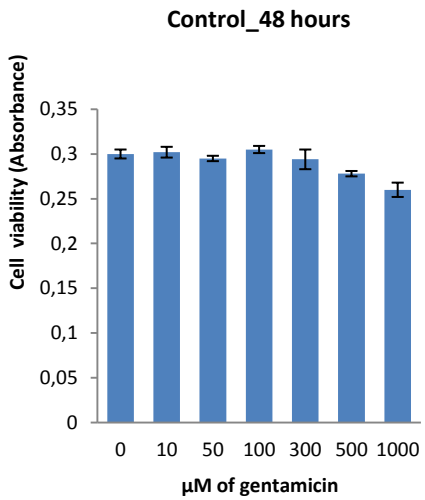
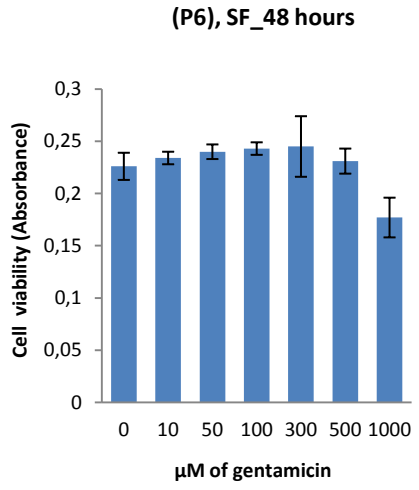
Patient 6

Control

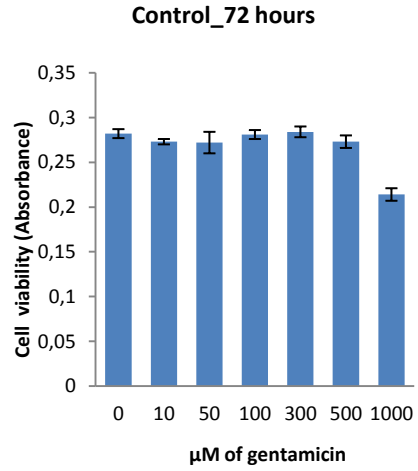
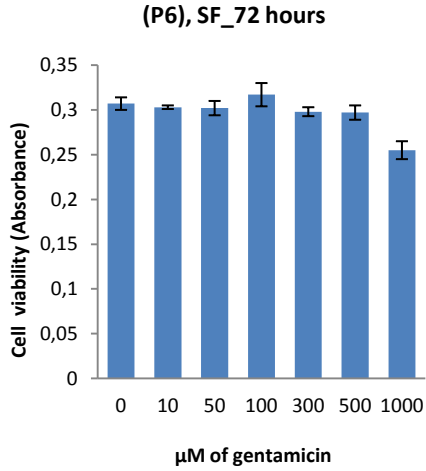
24 hours



48 hours



72 hours



E

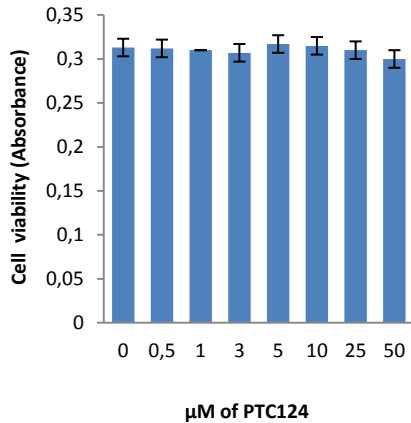
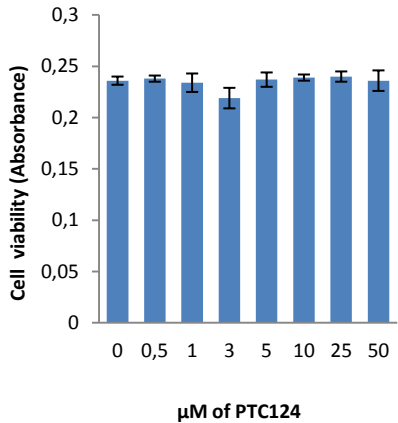
Patient 6

Control

16 hours

(P6), SF_16 hours

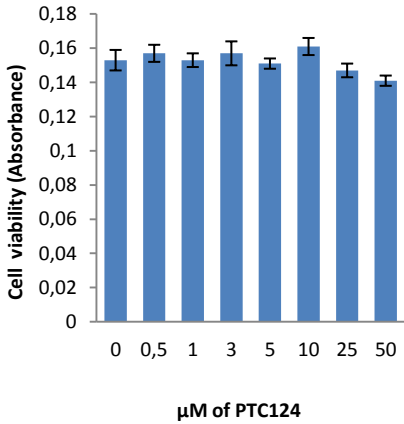
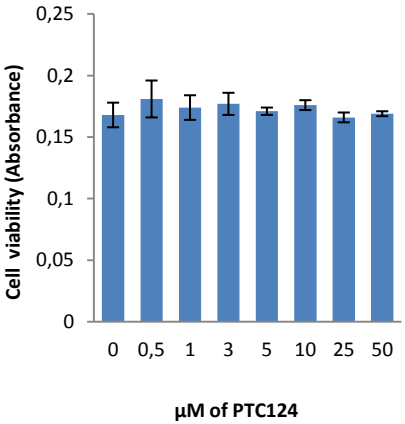
Control_16 hours



24 hours

(P6), SF_24 hours

Control_24 hours



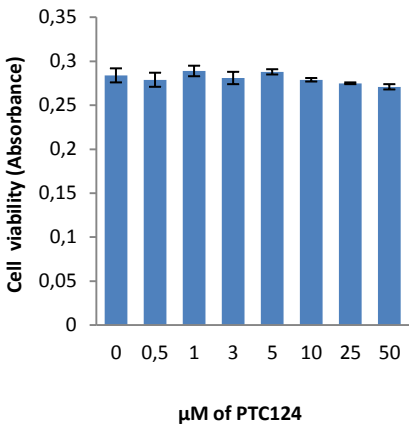
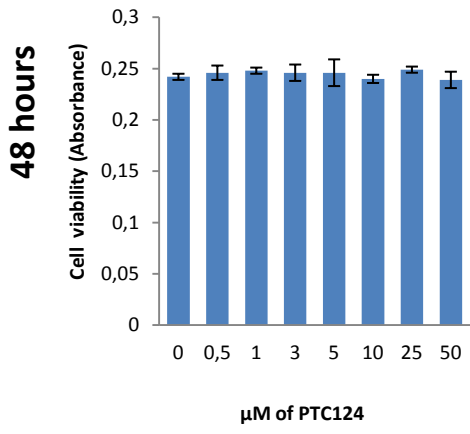
F

Patient 6

Control

(P6), SF_48 hours

Control_48 hours



(P6), SF_72 hours

Control_72 hours

