PALB2 mutations and the clinical management of breast cancer patients with breast cancer was identified to be ~5%.

PALB2 mutation prevalence in a large US referral population was 0.81% overall, with the Higher Risk group at 1.05% (95% C.I., 0.5-1.92) and the Lower Risk group at 0.38% (95% C.I., 0.05-1.37). This difference between the two groups was not statistically significant at the 5% level.

The overall VUS rate of PALB2 in this patient population with breast cancer was identified to be ~5%.

Conclusions: PALB2 mutation carriers currently exist but increased breast surveillance or participation in research studies may be appropriate.

Discussion: The positive rate is about 2.74 times higher in the Higher Risk group compared to the Lower Risk group. These results challenge the current 5% breast cancer rate in this population. Using an exact Fisher test, we obtained a one-sided p-value of 0.018.

The mutation prevalence of PALB2 in this population of breast cancer patients, negative for BRCA1 and BRCA2 mutations was 0.81% overall, with the Higher Risk group at 1.05% (95% C.I., 0.5-1.92) and the Lower Risk group at 0.38% (95% C.I., 0.05-1.37). This difference between the two groups was not statistically significant at the 5% level.

The overall VUS rate of PALB2 in this patient population with breast cancer was identified to be ~5%.