

## BRCA1 and BRCA2 protein expressions in an ovotestis of a 46, XX true hermaphrodite

### ABSTRACT

Gene expressions of breast cancer susceptibility genes such as BRCA1 and BCRA2 encode complex multi-factorial cellular functions, and mutation at the germ line predisposes individuals to breast and ovarian cancer but there is still strong evidence linking these genes to homologous recombination (through decomposition of DNA) and transcriptional control; also to tissue proliferation. BRCA1 and BCRA2 protein distribution in an unsusceptible tissue (an ovotestis) is the focus of our investigation, as there is disagreement over their localization to either nucleus or cytoplasm and whether their expression is present in premeiotic germ cells or can still be expressed in mitotic spermatogonia.

### INTRODUCTION

Three homologous members make up the Fragile X Mental Retardation (FMR) protein family, with the FMRP being encoded by the x-linked FMR1 gene and its absence is linked to human hereditary mental retardation [reviewed in]. This family includes two other members: the Fragile X Related 1 (FXR1P) protein, coded in human by the FX2P gene located at 3q28 and 17p13.1, and another lineage that encodes two KH domains and a RGG box—notes in functional characteristic motifs of RNA-binding proteins. They also contain a nuclear localization signal (NLS) and NES, which make them putative nucleocytoplasmic shuttling proteins (reviewed in ). There are indications that their functions may be related to RNA transport and/or translation. While FMRP is the cause of Fragile X Mental Retardation in human, it is unclear whether FXR1P and FxR2P are associated with any pathology or phenotype. Additionally, there is no evidence that these homologous proteins can counteract this absence. In vivo studies revealed that all three members interact with each other and in the same way. However, their expression patterns showed distinct patterns in certain tissues of mice and humans, suggesting that each protein may also function independently. Six different isoforms were identified and their levels were found to be specific to each cell type, indicating that FXR1P displays a complex expression pattern in various mammalian cell lines. They found four different FXR1P isoforms of MW 70 and 74 (previously called short) and later 78 and 80 kDa (long) are widely expressed in various cell lines as well as in mouse organs. The replacement of isoforms with novel MW 82 and 84 kDa super long isomerisms occurs in muscle. This phenomenon is evident during myogenesis of myocyte cell lines that can differentiate into myotubes. The model system that replicates, albeit imperfectly, muscle differentiation has enabled us to demonstrate, as previously mentioned, in the present report, that the short and long isoforms undergo a transitional event that coincides with the expression of muscle-specific genes, leading to the super long transition. Furthermore, we demonstrate that super long isoforms are expressed in low levels by undifferentiated myoblasts and are stored in the nuclei, whereas in different myotubes, P82,84 are transferred to the cytoplasm and become part of mRNPs found in actively translating ribosomes.

### CONCLUSION

It can be inferred that in Thai HIV-infected patients, intestinal parasite infections are still highly prevalent, both opportunistic and non-obsessed. Spira analysis is still a useful method of examination for HIV infection

among patients in Thailand who have either diarrhea or no symptom of the disease. As previously reported, suspected opportunistic intestinal parasite infection should be suspected in all patients with low immunity and HIV infection who present with diarrhea. The importance of tropical epidemic non-opportunistic intestinal colony infections should not be ignored.