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BRCA1* mutation spectrum, functions and therapeutic strategies: The story so far...**Babita Sharma¹, Raman Preet Kaur¹, Sonali Raut,¹ and Anjana Munshi^{1*}¹Centre for Human Genetics and Molecular Medicine, Central University of Punjab Bathinda, Punjab, India Corresponding Author Address**

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BRCA1* functions, mutation spectrum, and therapeutic strategies: The story so far...*1. Introduction**

Breast Cancer is not only one of the most commonly diagnosed cancer (23%) but also a leading cause of cancer-related deaths (14%) among women world over [1]. In 2015, WHO estimated 231,840 new cases of invasive breast cancer and 60,290 additional *in situ* cases. The incidence rate varies drastically across the world, ranging from 92 per 100,000 in North America to 27 per 100,000 in Middle Africa and Eastern Asia [1]. The Ashkenazi population represents the Jews having ancestors from Eastern and Central Europe, such as Germany, Poland, Lithuania, Ukraine, and Russia [2, 3]. The hereditary proportion contributing to breast cancer in Ashkenazi population is much higher in comparison to other ethnic groups on account of the founder mutations [3, 4]. In the developing countries, like Asia and Africa, the disease is increasing at an alarming rate because of development and modernization mainly due to the sedentary lifestyle and increasing consumption of liquor [5]. Poverty, illiteracy, the social hitch of women and lack of proper/in time medical facilities are some of the causative factors for its late diagnosis contributing further to the drastically increasing rate of incidence in these regions [5].

Genetics plays a pivotal role in augmenting the risk in the family having a history of breast cancer via some inherited genetic mutations [6]. The most susceptible cause for breast cancer is due to high penetrant genes like *BRCA1* and *BRCA2* genes and a number of low penetrant genes including Ataxia-Telangiectasia Mutated (*ATM*), Checkpoint kinase 2 (*CHEK2*), Partner and localizer of *BRCA2* (*PALB2*), *BRCA1* Interacting Protein C-terminal Helicase1 (*BRIP1*), Protein 53 (*P53*), Phosphatase and Tensin Homolog (*PTEN*) and Cadherin-1(*CDH1*) [7]. All these genes play an integral role in multiple signaling pathways inside the cell. The crosstalk between these proteins maintains a delicate balance for proper functionality of cellular machinery. When one of these signaling molecules become non-functional, this balance gets disturbed, which may assist in the progression of carcinogenesis.

BRCA1 is one of the well-studied genes associated with breast cancer. There are more than 13,000 research articles related to *BRCA1* gene in PubMed with more than 2200 articles related to *BRCA1* gene function. Women with the mutated

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