



Risk Assessment and Psychological Implications of Genetic Counselling

**Pooja Sethu Madhavan^{1#}, Jennifer Sally Samson¹⁺, Sushmashri Chelliah^{1@},
Vanishree Venkatasubramaniam^{1S} and Venkatachalam Deepa Parvathi^{2*}**

*¹Department of Human Genetics, ²College of Biomedical Sciences, Technology and Research,
Sri Ramachandra Institute of Higher Education and Research Porur, Chennai 600 116,
Tamil Nadu, India*

*E-mail: #<poojasm95@gmail.com>, +<sallysamson93@gmail.com>
@<coolsushma15@gmail.com>, <vani1298.venkat@gmail.com>*

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ABSTRACT Genetic diseases and diagnostics have now become a part of mainstream medicine. This has led to the augmentation of genetic counselling into routine medical practice as and when the need arises. The elements of genetic counselling include examination of family history, deconstructing the disease and its manifestation, risk assessment and understanding the available management options. In this review paper, the authors have extensively examined the factors involved in the risk assessment such as Mendelian inheritance, age, consanguinity and other complexities such as *denovo* mutations, penetrance, germ line mosaicism, variable expression, polygenic inheritance, multifactorial inheritance, uniparental disomy and genomic imprinting. The technological advancements that have eased the process of risk calculation have been explored. Genetic counselling often acts as a harbinger of bad news, albeit unintentionally. This leads to undue pressure on the consultant, leaving psychological implications. Hence the psychological implications of genetic counselling have been detailed.