5. CONCLUSION

Parkinson’s disease (PD) is a progressive degenerative disorder of brain caused due to the death of dopaminergic neurons in Substantia Nigra. The cardinal manifestations of PD are resting tremor, rigidity and unstable gait.

In India, approximately 0.32 million individuals suffer from PD, whereas it is reported that approximately 1.2 million individuals suffer from PD in western population. The alarming rise in the prevalence of PD in India has been attributed to the demographic pattern, changing environment, as well as lifestyle. In Karnataka, the prevalence of PD is reported to be 7 per 100,000. In the later studies, the prevalence of PD above 50 years of age was 134 per 100,000. This indicates the spontaneous increase in the incidence of PD in elder population (142).

Our study is the first of its kind in North Karnataka, which explored the role of possible genes involved in Parkinson’s disease with cutting edge technologies like Next Generation Sequencing (NGS). We analysed the role of SNCA gene in Parkinson’s disease. The absence of SNCA gene mutations in Parkinson’s disease in this population prompted us to target other possible genes which may be playing role in the etiology of Parkinson’s disease in this population. Hence, we subjected some PD samples for NGS. Through NGS, we have identified the NOS1 gene present on chromosome number 12 to be involved in Parkinson’s disease in this population. Identification of this gene has opened new vistas to the clinicians and biologists to target Parkinson’s disease in this population specifically in terms of molecular analysis of NOS1 gene.

In the present study, on chromosome number 12, we found 7bp novel insertion +GGTAAAA at position 117718473 in NOS1 gene. This is the first report which explored the possibility of novel insertions in Parkinson’s disease. According to our knowledge screening for NOS1gene mutations or insertions for early diagnosis of Parkinson’s disease in this population may have some significant meaning, but with the small sample size, it may not be possible to elucidate the exact role of NOS1 gene in Parkinson’s disease. Hence, it is essential to screen more number of samples and other risk factors which are predicted to play role in development of Parkinson’s disease.

PD is a neurological disorder in which many gene pathways and genes are involved. To understand the exact etiology and molecular mechanism of PD it is
necessary to carry out **Meta analysis based on exome sequencing** with the more number of samples in different populations.

India is known for high degree of inbreeding with its heterogynous population. This makes it necessary to screen a large number of patients perhaps within each group in order to get a true picture of contribution of genes in the etiology of PD. In order to find out the prevalence of any specific mutations, a large number of families need to be investigated.