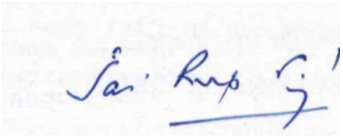
g. Citation (summary) on the outstanding research work

The applicant has contributed immensely to identify the genes involved in ischemic stroke (IS) and its subtypes in the Indian population. The genes evaluated include ACE, PDE4D, eNOS, CYP11b2, E-selectin, estrogen receptor α, IL-10, MMP3, CYP4F2, LPL genes (34, 41, 56, 57, 59, 65, 73, 76, 77, 80, 81, 86). The pharmacogenomics of statins and aspirin has been evaluated at length suggesting personalized medicine options for these patients (51, 63). The role of serum uric acid, serum albumin & C-reactive protein has also been evaluated in IS and its subtypes (50, 52, 64). The platelet parameters and the genes (ARHGEF3 & THPO) along with their expression affecting the MPV have been established to be associated with the development of IS and its subtypes (4).

The mutations associated with thalassemia and Sickle Cell Anemia involving 5000 patients have been delineated. This has helped in providing counselling to the parents of the children affected by these diseases. The role of modifier genes like BCL11A, Xmn1 restriction site polymorphism has been evaluated & confirmed that increased γG-globin expression ameliorates the clinical severity of β-thalassemia (31, 45). Further, the genes (UGT1A*2) involved in pharmacogenomics of deferiprone in β-thalassemia major have also been identified (47).

The common mutations associated with breast cancer in Malwa region of Punjab including the novel mutations in male breast cancer have been established (23, 13). In addition, the elevated levels of IL-6 & IL-17a associated with poor outcomes in patients and can be used as prognostic markers (20).



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