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PRSS1 GENE MUTATION IS RARE IN INDIAN PATIENTS WITH HEREDITARY PANCREATITIS WHO HAVE DISEASE TRAJECTORY SIMILAR TO SPORADIC CP WITH SPINK1 MUTATION

**Society:** AGA**Track:** Pancreatic Diseases**Author(s) and Affiliation(s):**

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**Introduction:** Hereditary pancreatitis (HP) is an autosomal dominant disorder that is mostly caused by cationic trypsinogen gene (*PRSS1*) mutation in the Caucasian population. The prevalence of *PRSS1* gene mutation in HP is 70-80% in Europe and USA, and 40% in Japan. Our objective was to study the mutational profile of patients with HP in India and compare them with mutation-matched sporadic CP patients.

**Methods:** Patients with chronic pancreatitis (CP) were recruited from January 2000 till July 2024. The diagnosis of HP was made on the basis of presence of pancreatitis in two first degree relatives or three second degree relatives. Genetic mutations in 5 genes i.e. *PRSS1*, *CFTR*, *SPINK1*, chymotrypsin C (*CTRC*) and carboxypeptidase A1 (*CPA1*) were tested by sequencing all the 5 genes. Clinical characteristics, pain pattern and need for endoscopic or surgical intervention of *SPINK1* mutation positive HP patients was compared with *SPINK1* mutation positive sporadic CP.

**Results:** A total of 83 participants from 34 families were included (49 symptomatic with CP, 60.2% male, alcoholism 10%, smoking 6%). *PRSS1* mutation (N29I) was detected only in one family. *SPINK1* gene mutation was the commonest in 25 families (p.N34S in 23 families, c.194+2T>C and c.-215G>A in one each), *CFTR* mutation in 4 families, *CTRC* in 5 families and *CPA1* in two families (Table). The median (range) age at onset of symptoms of patients with HP was 19 (3-47) years and at diagnosis 23 (7-58) years. Clinical presentation was recurrent acute pancreatitis in 75.5% patients and the remaining had CP at first presentation. 49% required endoscopic or surgical intervention over a median follow up of 11 (2-24) years for pain relief (Figure). 33% of patients developed diabetes after a median of 7 years. During 633 person-years of follow up, 3 patients died including 2 due to complication of HP. On comparing *SPINK1*+ HP (n=39) and *SPINK1*+ sporadic CP (n=69) during the study period, no difference was found in the age of presentation, pain pattern or need for intervention.

**Conclusion:** Unlike in the Caucasian population, *PRSS1* gene mutation was rare in Indian patients with HP. The most common mutation in our Indian cohort was *SPINK1* mutation. The disease onset and clinical trajectory of *SPINK1*+ HP were similar to *SPINK1*+ sporadic CP. A deeper insight into population genetics is required to understand the differences between Indian and Caucasian patients with HP.

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