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Epidemiology and molecular biology of gastrointestinal stromal tumors (GISTs): a population-based study in the South of Switzerland, 1999-2005.

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Introduction. Gastrointestinal stromal tumors (GISTs) are characterized at the molecular level by c-kit or PDGFRA oncogene mutations. Although GISTs raised major interest in past decades, population-based studies are still rare.

Materials and Methods. All GISTs diagnosed in Southern Switzerland (1999-2005) were identified using Ticino Cancer Registry and analysed for c-kit and PDGFRA mutations. Clinical and molecular features were studied.

Results. Annual incidence of GISTs was 1.47 cases/100,000 inhabitants (median age: 64 years; median size: 6.0 cm). Most GISTs arose in the stomach (60.5%). The malignancy risk was very-low/low in 47% of patients. DNA sequences showed a gene alteration in either c-kit or PDGFRA genes in 72.5% of patients. Mutations occurred mostly in c-kit exon 11 (60%). No mutations in c-kit exons 13 or 17 were found. An equal number of alterations in exons 12 and 18, and no mutations in exon 14 were observed in the PDGFRA gene.

Discussion. This is the first comprehensive population-based study of GISTs incidence and molecular biology characterization in Central Europe. Our incidence data showed higher age-standardized rates compared to other European countries. The gene mutation spectrum differed when compared to the literature. This is relevant to improve the molecular profile knowledge based on Cancer Registry data.