# Spectrum of MLH1 and MSH2 mutations in Chilean Families with suspected Lynch syndrome

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PURPOSE: Lynch syndrome is the most common inherited syndrome of colorectal cancer, caused principally by germline mutations in MLH1 and MSH2. We report our experience with genetic screening in the diagnosis of Lynch syndrome in Chile, a country previously underserved in the capacity to diagnose hereditary colorectal cancer. METHODS: Families from our Familial Colorectal Cancer Registry were selected for this study if they fulfilled either Amsterdam I/II or Bethesda criteria for classification of Lynch syndrome. Analysis of colorectal tumors from probands included a microsatellite instability study and immunohistochemical evaluation for MLH1 and MSH2. Screening of germline mutations was performed by single-strand conformation polymorphism analysis and DNA sequencing. RESULTS: A total of 21 families were evaluated, 14 meeting Amsterdam criteria and 7 meeting Bethesda criteria. Tumors in 20 families (95%) showed microsatellite instability (19 high and 1 low) and 9 of these 20 families (4