INTRODUCTION: Patients with Gaucher disease (GD) are considered to be at an increased risk of malignant disorders. Much of the observation reported to date is based primarily on European or North American studies, but no studies have addressed this issue in South America. OBJECTIVES: Evaluate the incidence and characteristics of malignant diseases in a cohort of Argentine patients with GD type 1, and direct relatives (parents and siblings) during their follow up. METHODS: Medical doctors in charge of Gaucher patients, from all around the country, mostly hematologist and pediatricians, were requested to report patients and direct relatives with malignancies confirmed by histopathological study. A form was sent, without patient identification. RESULTS: Out of a total of 124 patients, one event of cancer was reported in 15 of them, and 2 patients had 2 events. All patients were receiving enzyme replacement therapy (ERT), none were splenectomized. The mean age of the patients that developed CD was 51 years (range: 27-68) 8 females, 7 males. CD reported were: colon (3), kidney (2), breast, lung, ovary, hepatocellular carcinoma, prostate, testicle, rectum, mieloma, Non Hodkin Lymphoma (NHL) and retroperitoneal sarcoma (1 case each). Progression of the CD was presented in 60 % and 40 % died as a consequence of the CD after 2.5+-1.5 years of diagnosis. Time between GD and CD diagnosis: 0-28 years, 2 cases simultaneous. GD patients direct relatives with CD: 8 cases. Mother 4 cases: uterus, ovary, breast and colon. Father 1 case: colon. Brothers 3 cases: pancreas, ovary, breast and thyroid. 2 families jew ashkenazi (370/84 GG) with multiple cancer. Family 1: mother breast and colon, sister: breast and thyroid. Family 2: mother, maternal aunt and grandmother: breast. CONCLUSIONS: In a population of 124 patients with GD type 1 from Argentina the search of malignancies was performed. A total of 15 patients with malignancies was found, 2 patients presented 2 neoplasms: kidney and colon and breast and ovary. In direct relatives 8 cases were found and 2 families presented multiple cancers. No pediatric case were reported.