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Short bowel syndrome is a rare disease gaining popularity

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The National Organization of Rare Disorders states that Short Bowel Syndrome (SBS) affects males and females in equal numbers. The disorder is usually acquired during life but in rare cases may be present at birth (congenital). In adults, short bowel syndrome usually results from the surgical removal of a portion of the small intestine. Crohn's disease is the most frequent cause of surgical removal of the small intestine in adults. In newborns, necrotizing enterocolitis is the most common cause of surgical removal of the small intestines. The exact incidence and prevalence of short bowel syndrome in the general population is unknown. Before 2010, SBS was a condition with very little support for its patient base. In 2016 the support is very strong with a large patient and family population base and has taken interest by the pharmaceutical industry that is innovating new therapies for the rare disorder. What changed? How did support go from almost nobody to a population of active 3000 members and growing? Foundation support was the answer by many different patient organizations that saw the need to support SBS as a known condition; the congenital defect causing it and how to live a better quality of life. SBS is now a very rare disorder that is known by many, but still studied by few.

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RAS/BRAF mutational status in familial non-medullary thyroid carcinomas

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There are contrasting views on whether familial non medullary thyroid carcinomas (FNMTCs) are character¬ized by aggressive behavior and limited evidence exists on the prognostic value of *BRAF* and *RAS* mutations in these tumors. Thus, in the present study, clinic pathological features were analyzed in 386 non medullary thyroid carcinomas (NMTCs), subdivided in 82 familial and 304 sporadic cases. Furthermore, the *RAS* and *BRAF* mutational statuses were investigated in a subgroup of 34 FNMTCs to address their clinical and biolog¬ical significance. The results demonstrated that, compared with sporadic NMTCs, FNMTCs are characterized by significantly higher rates of multi-centricity and bi-laterality and are more frequently associated with chronic autoimmune thyroiditis. Notably, a statistically significant difference in the rates of multi-centricity was observed by sub-grouping familial tumors according to the number of relatives involved; those with ≥3 affected relatives were more likely to be multi-centric. Further¬more, the FNMTC cohort exhibited higher rates of tumors >4 cm in size with extra-thyroidal or lymph node involvement. However, no significant difference was observed. Similarly, no differences were observed with respect to the age of onset or the patient outcome. The mutational profiling exhibited a rate of 58.8% for BRAF V600E mutations in familial tumors, which is at the upper limit of the mutational frequency observed in historical series of sporadic thyroid cancer. A high rate of NRAS mutations (17.6%) was also observed, mostly in the follicular variant histo-type. Notably, compared with BRAF/RAS wild-type FNMTCs, the familial carcinomas bearing *BRAF or NRAS* mutations exhibited slightly higher rates of bi-laterality and multi-centricity, in addition to increased frequency of locally advanced stage or lymph node involve¬ment. The present data support the theory that FNMTCs are characterized by clinic-pathological features that resemble a more aggressive phenotype and suggest that *RAS/BRAF* muta¬tional analysis deserves to be further evaluated as a tool for the identification of FNMTCs with a potentially unfavorable prognosis.

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