Abstract (lay version) of project

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Next generation sequencing approach to desmoid tumors

Desmoid tumors are proliferations of relatively benign appearing fibroblasts. Despite their histologic bland appearance, a significant subset of these tumors recurs aggressively and requires often debilitating surgery. Currently there are no molecular markers that predict the behavior of desmoid tumors. The purpose of this study will be to perform a very broad molecular search for markers that can be used to address two clinically highly relevant questions.

First, it is well known that not all desmoid tumors behave in the same manner. Some are very aggressive, others have an indolent behavior. While attempts have been made to develop predictors for the behavior of desmoids, these rely only on clinical parameters such as tumor size, site and age of the patient and no molecular markers for recurrence risk have been identified. At this moment it is impossible to tell with certainty which tumors require aggressive treatment and which can be followed by “watchful waiting”. Through our ability to perform next generation sequencing on archival tumor samples we can now perform a very broad search for changes in the genetic material that can help us predict the behavior of desmoids. In addition to identifying markers that can be used to predict the behavior of desmoid tumors, we will also perform an in-depth search for markers that can be used by pathologists to distinguish scar from desmoid recurrence. The data that we will acquire under this proposal will complement the already existing dataset in our laboratory.

We have previously performed quantitative measurement of the expression levels for all know human protein-encoding genes in 9 desmoid tumor samples, 4 scars and 42 non-desmoid fibroblastic lesions. The number of scar samples that we have analyzed was too low but the preliminary results were encouraging. Our current dataset includes 29 primary, 20 recurrent desmoid tumors and 15 scars from 26 patients.