Grassroots effort to open hereditary disease research to the public

Genes exist as interrupted fragments that get joined together by a process called splicing.

One in three hereditary disease alleles affect the splicing of genes. This may add to the severity of the disease.

There are examples of drugs and oligo therapies can reverse these splicing defects. This may reduce the severity of the disease.

Our goals:

1) Offer a public and anonymous website to allow individuals to predict which mutations affect splicing (http://fairbrother.biomed.brown.edu/spliceman/index.cgi)

2) Develop a high throughput free and anonymous testing service that a) determines if a mutation changes splicing and b) tests the effect of several existing FDA approved compounds on the processing of that mutated gene.
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Experimental design:
The experiment requires the sequence of the piece of DNA around the mutation and the mutation itself. The user could input this information into an online form. This information is used to synthesize the gene and test its behavior in tissue culture cells. The experiments are done in batches of at least 6,000. It would take several months to fill a batch and do the experiment. We could email the result back to the user. Successful drug treatments might be used to initiate a clinical trial.

Questions:
1) Would there be interest in using this resource?

2) Could users become familiar enough with genomics and biology to enter the location and sequence of their mutation into an online form?

3) Would email contact be sufficient to preserve anonymity but stable enough to allow us to keep in touch in the event a clinical trial is organized down the road?

Comments, Ideas?
Prof. Will Fairbrother, MCB Dept Brown University  
fairbrother@brown.edu  
401 863 6215