Hi Phil,
We finished the mutational analysis. Please note that these results are for research purposes only and do not constitute an official diagnosis. We found that NM is a compound heterozygote for two mutations, a 3556 G to C splice junction mutation and an Arg127Gln mutation. The splice junction mutation is a common mutation and, as I see from your web site, was found at MGH (they use a slightly different numbering system based on our initial call of the gene sequence- our current numbering reflects some subsequent revisions).