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Autosomal recessive disorders are disorders that can arise ina child whose parent show no symptoms of that condition.

How is this possible? It’s possible because the parents are “ carriers” of only one copy of theabnormal gene, instead of two. In the slight chance that both parents pass ontheir abnormal gene to their child, the child now has both copies of theabnormal gene, and will therefore show the symptoms of the disorder associatedwith that abnormal gene. This information allows many prospective parents tothink, “ Is carrier testing worth my time and money?” Niemann Pick Disease (NPD) is an example of an autosomalrecessive disorder. NPD causes a build-up of lipids (fat) in the cells ofvarious organs. NP can be divided into 4 types that differ slightly intheir severity and symptoms. Here, we will focus on the rare and chronic formof NPD; Type C. As a brief overview of NPD-C; an individual with NPD-Cinherits the 2 mutated copies of the “ NPC1″ or NPC2” gene from their mother andfather.

Current research suggests that the NPC genes codes for proteins thatare involved in the transport of lipid molecules in the cells. Mutated NPCgenes therefore results in the inactivity of these necessary transport proteins; leading to a buildup of lipids, such as cholesterol in the cells. Ultimately, NPD-C results in tissue and organ damage; specifically, in the spleen, liver, and/or lungs. There are also neurological complications associated with NPD-C. Somesymptoms of NPD-C include seizures, difficulty moving limbs (dystonia), liverdisease, and difficulty with speech. There is currently no cure for NPD-C, however, there are treatments for the symptoms; such as medications called “ Miglustat”, and “ Clomipramine”; psychosocial support

for the child and family; andphysical/speech/occupational therapy. NPD-C is estimated to affect 1 in every100000 individuals, and it is most common in Nova Scotian people with FrenchAcadian descent.

It is predominantly prevalent during childhood, with someindividuals surviving up to adulthood.  Two tests associated with NPD-C are skin assays and genetictests. Skin assays are diagnostic tests; where a small skin sample is used toexamine how the cells transport and store cholesterol. The results of this testare good indicators as to whether or not an individual has NPD-C. Alternatively, a genetic test is often used to determine if a parent is acarrier for the focal disease. As mentioned earlier, NPD-C is an autosomalrecessive disorder.

Meaning both parents must be carriers of an abnormal genein order for there to be a 25% chance of their future child inheriting thedisease. Genetic testing is a common subject for debate, about whether or notit is beneficial. One disadvantage of genetic testing for NPD-C is the fact thegenetic test can only determine if an individual is a carrier is if theabnormal gene is identified and described; based on the first person in thefamily to have the disease. This is a major limitation of genetic testingbecause it would mean that multiple family members that have the disease mustbe tested. Not only is this time- consuming, but when multiple family members requiretesting, this can also be very costly (around hundreds of dollars), which isanother disadvantage of genetic testing. Although there are a few limitations and disadvantages togenetic testing for NPD-C, for prospective parents who have a family history ofNPD, the pros definitely outweigh the cons.

Firstly, if the genetic testresults show that both or one of the prospective parents are not carriers forthe mutated NPD-C gene; this alleviates a lot of unnecessary stress and anxietysurrounding the situation for the parents. However, even if the test resultsshow that both parents are carriers, this information allows parents to preparethemselves. The reality is, that there is a small chance that their future childmay inherit NPD-C. By taking the genetic test before becoming pregnant, theseprospective parents are giving themselves time to talk to a genetic counselor, and understand the risks involved with having a child, and what their otheroptions are for family planning (artificial insemination, sperm donor, etc.). Ultimately, genetic tests are extremely valuable for all prospective parentswho has a family history of NPD-C, no matter what the results of the test are.  In conclusion, if you and your partner have a familyhistory of NPD-C, and are planning to start a family, then the short answer isyes... genetic testing is worth yourtime and money.