Date

Mailing Address

Dear :

We received your consent form to receive genetic test results. You indicated that you wanted research test results for any monogenic (single gene) disease that we studied. Therefore, this letter includes results for 14 genetic conditions that have been identified in the South Dakota Hutterites and that we have tested for.

All genetic conditions are caused by changes in our genes. Genes are the inherited instructions that guide the growth, development, and function of our bodies. Everyone has two copies of most genes. Each of the 14 conditions we tested for is associated with a different gene. Individuals affected with these conditions have genetic changes (called mutations) in both copies of the gene associated with that specific condition. There are some conditions, however, where an individual may not ever develop symptoms even if they have genetic changes in both copies of the gene for that condition.

It is possible that you are a carrier for at least one of these conditions. *Carriers* for these conditions have a change in only one copy of their gene. Because their other gene is unchanged, carriers are healthy and typically have no signs of the disease. If your spouse (or future spouse) is a carrier for the SAME condition, each of your children has a 1 in 4 (25%) chance of inheriting the change from both parents and having that condition, and each of your children has a 1 in 2 chance (50%) of also being a carrier.

If only one parent is a carrier for a condition and the other is not, the risk is nearly zero that you will have a child with that condition. There is a 50% chance of having a child who is a carrier, like the parent. If you are a carrier for a particular condition, your siblings could be carriers (or affected) with that condition.

If you receive a negative result for one of these conditions, the chance you are still a carrier is small, although it is not zero. Some people may have a mutation the test does not look for, however this is unlikely. In addition, we only test for the genetic changes that are known to cause these 14 conditions in the Hutterites and you could still be a carrier for mutation(s) causing any of the remaining recessive conditions in the Hutterites.

Please note that these results are considered “research” and should not be directly used in guiding your care. Genetic results from a research laboratory could be in error. Therefore, you should have the results of this research confirmed in a clinical laboratory if they are needed to guide your medical care or decision making. Your doctor should be able to help you arrange this clinical testing and provide counseling about the results. If

your doctor is not sure how to arrange clinical testing, we can help you with this if you desire.

I have included a copy of your signed consent form. If you have any questions about these results, please contact me by mail or phone at (XXX) XXX-XXXX. Results are available for most adults (18 years of age or older) who participated in our previous studies if they are interested. I hope this information is helpful to you and I thank you for participating in our studies.

Sincerely,

Genetic research results for NAME (DOB):

Name of condition	Gene name	RESULTS*
Arrhythmogenic right ventricular cardiomyopathy (ARVC)	*DSC2*	
Bardet-Biedl syndrome	*BBS2*	
Cystic fibrosis	*CFTR*	
Dilated cardiomyopathy with ataxia	*DNAJC19*	
Joubert syndrome	*TMEM237*	
Limb girdle muscular dystrophy 2H	*TRIM32*	
Limb girdle muscular dystrophy 2I	*FKRP*	
Nonsyndromic deafness	*GJB2*	
Oculocutaneous albinism type 1	*TYR*	
Restrictive dermopathy	*ZMPSTE24*	
Sitosterolemia	*ABCG8*	
Spinal muscular atrophy type 3	*SMN1*	
TECR syndrome	*TECR*	
Usher syndrome type 1F	*PCDH15*	

*There is one specific genetic change (mutation) that has been previously identified in the Hutterites for each condition except cystic fibrosis, where there are two known mutations. Please contact us if information about the specific genetic change(s) is needed for medical care.

Definitions of Results

Not a carrier: Neither copy of this gene has the Hutterite mutation

Carrier: One copy of this gene has the Hutterite mutation

Affected: Both copies of this gene have the Hutterite mutation; these individuals may or may not have or ever develop symptoms depending on the condition

Unknown: No results available for this condition. This could be due to technical issues or limited amounts of your DNA sample in the lab.

Descriptions of these conditions are included in the brochure you received previously. Please contact us if you would like additional information on any of these conditions.