

# GENETIC TEST SUMMARY

Donor: 14114

ANCESTRY

English, Welsh

JEWISH ANCESTRY?\*

No

IT IS STRONGLY RECOMMENDED THAT YOU DISCUSS THE DONOR'S RESULTS WITH YOUR PHYSICIAN PRIOR TO SHIPMENT TO VERIFY THAT THIS DONOR IS SUITABLE FOR YOUR USE.

## TEST RESULTS

GENETIC TEST	RESULT	DETAILS / ESTIMATED RESIDUAL RISK**
Chromosome (karyotype) analysis	Normal male karyotype	No evidence of a clinically significant chromosome abnormality
Hemoglobin evaluation	Normal hemoglobin fractionation and MCV/MCH results	Reduced risk to be a carrier for sickle cell anemia, thalassemias, and other hemoglobinopathies
Cystic fibrosis carrier screening	Negative by targeted mutation analysis	1/420
Spinal muscular atrophy carrier screening	Negative for deletions(s) of exon 7 of the SMN1 gene	1/834
Herlitz Junctional Epidermolysis Bullosa	Negative by selective sequencing of the LAMC2 gene	1 in 666
Glutaric Acidemia, Type I (GCDH)	Negative by sequencing analysis of the GCDH gene	1/1,900
Lamellar Ichthyosis, Type 1 (TGM1)	Negative by sequencing analysis of the TGM1 gene	1/1,100
Methylmalonic Aciduria/Homocystinuria: type cbIC (MMACHC)	Negative by sequencing analysis of the MMACHC gene	1/6,800

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This donor's GTS was originally created: 06/09/15 and last revised: 08/28/19.  
Results are subject to change without notification.

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**ALL people carry genetic mutations for disorders inherited in an autosomal recessive (AR) manner, and some of these mutations can be detected by genetic carrier screening. A person's offspring are not expected to develop that condition unless they inherit mutations for the same AR condition from BOTH parents.** Therefore, CCB strongly recommends that all recipients and their physicians discuss a donor's genetic test results PRIOR to shipment of a donor's specimens, to ensure that the results are suitable for the recipient's reproductive plans. CCB also recommends that the recipient meet with a genetic counselor who can help to explain the donor's results and testing options that may be appropriate for the recipient to consider.

**Genetic testing can only reduce the chance for specific inherited conditions in a donor's offspring; it cannot eliminate the risks for those specific disorders or other untested conditions.** There is always a 3 to 4% chance to have a child with a medical issue, regardless of the screening performed.

\*Please see the Donor Profile for details on the type of Jewish ancestry (Ashkenazi, Sephardic, maternal, paternal, etc.).

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