



Reproductive Technologies, Inc.

# THE SPERM BANK OF CALIFORNIA

## EXPANDED CARRIER SCREENING RESULTS DONOR 6104

Expanded carrier screening for 525 autosomal recessive conditions was completed by Invitae and reported on 04/11/2023.

The results were **NEGATIVE** for all **pathogenic recessive conditions**. Donor 6104 is a **POSSIBLE carrier for Congenital nephrotic syndrome type 2 (NPHS2)**. The specific variant the donor carries may be pathogenic when combined with certain NPHS2 variants. Research on this variant is ongoing and information may change over time. **Recipients using this donor can consider carrier screening for the NPHS2 gene to further clarify risks.**

Testing was negative for the remainder of genes screened.

Disease	Result	Residual risk to be a carrier (based on **ethnicity** ethnicity)
<b>Congenital nephrotic syndrome type 2 (NPHS2)</b>	Possible carrier c.686G>A (p.Arg229Gln)	n/a
Cystic Fibrosis	Negative	1 in 4400
Spinal Muscular Atrophy	Negative: 3 copies	< 1 in 342
HBB Hemoglobinopathies & Thalassemia	Negative	1 in 48,00
Alpha Thalassemia	Negative	1 in 291

Genetic screening tests can significantly reduce, but never completely eliminate, the chance that a person is a carrier for a particular disorder.

Please refer to the donor's Invitae expanded carrier test report for more information on the testing completed and the donor's results.

**Please also see the Health Problems List for a summary of the information that this donor has provided to us regarding personal and family medical history.**

Sincerely,

Janine Mash  
LCGC Certified Genetic  
Counselor San Francisco  
Genetic Counseling