Midwest Sperm Bank

PATIENT RELEASE FOR DONOR GENETIC MATCHING

I, _____, have consulted with my physician concerning the use of **Donor** 687 and his genetic profile (see below). I understand that it is solely my responsibility to seek genetic counseling as well as genetic testing. By using donor _____ for purposes of therapeutic insemination/IVF, I hereby release Midwest Sperm Bank from all liability claims associated with the use of **Donor** 687.

Signature of Client: _____ Date: __/__/____

Signature of Medical Director: _____ Date: __/__/____

Donor # 687 is a carrier for the recessive disorder of

PHENYLALANINE HYDROXYLASE DEFICIENCY

which has a reproductive risk of 1 in 200.

21-HYDROXYLASE-DEFICIENT CONGENITAL ADRENAL HYPERPLASIA

which has a reproductive risk of 1 in 230.

STEROID-RESISTANT NEPHROTIC SYNDROME

which has a reproductive risk of 1 in 1600.

Counsyl

RESULTS RECIPIENT
MIDWEST SPERM BANK
Attn: Dr. Amos Madanes
4333 Main Street
Downers Grove, IL 60515
Phone: (630) 810-0217
Fax: (630) 810-0490
NPI: 1184790222
Report Date: 02/09/2018

MALE
AA 687
[redacted]
Ethnicity: Northern European
Sample Type: EDTA Blood
Date of Collection: 01/30/2018
Date Received: 02/01/2018
Date Tested: 02/09/2018
Barcode: 11004212318610
Indication: Egg or sperm donor

FEMALE
N/A

Foresight™ Carrier Screen

POSITIVE: CARRIER

ABOUT THIS TEST

The **Counsyl Foresight Carrier Screen** utilizes sequencing, maximizing coverage across all DNA regions tested, to help you learn about your chance to have a child with a genetic disease.

RESULTS SUMMARY

Risk Details	AA 687	Partner
Panel Information	Foresight Carrier Screen Universal Panel (175 conditions tested)	N/A
POSITIVE: CARRIER Phenylalanine Hydroxylase Deficiency **Reproductive Risk: 1 in 200** **Inheritance: Autosomal Recessive**	**CARRIER*** NM_000277.1(PAH):c.194T>C(I65T) heterozygote	The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group. Carrier testing should be considered. See "Next Steps".
POSITIVE: CARRIER 21-hydroxylase-deficient Congenital Adrenal Hyperplasia **Reproductive Risk: 1 in 230** **Inheritance: Autosomal Recessive**	**CARRIER*** NM_000500.7(CYP21A2):c.844G>T (V282L, aka V281L) heterozygote	The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group. Carrier testing should be considered. See "Next Steps".
POSITIVE: CARRIER Steroid-resistant Nephrotic Syndrome **Reproductive Risk: 1 in 1,600** **Inheritance: Autosomal Recessive**	**CARRIER*** NM_014625.2(NPHS2):c.686G>A (R229Q) heterozygote †	The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group. Carrier testing should be considered. See "Next Steps".

†Likely to have a negative impact on gene function.
*Carriers generally do not experience symptoms.

No disease-causing mutations were detected in any other gene tested. A complete list of all conditions tested can be found on page 11.

CLINICAL NOTES

- None

NEXT STEPS

- Carrier testing should be considered for the diseases specified above for the patient's partner, as both parents must be carriers before a child is at high risk of developing the disease.
- Genetic counseling is recommended and patients may wish to discuss any positive results with blood relatives, as there is an increased chance that they are also carriers.