

Name: Jane Doe

Donor: BFA0184

Consult Date: July 21, 2026

Indication

This note reviews carrier screening results for Sussex Sperm Bank Donor BFA0184.

Carrier Screening Summary

Donor BFA0184 completed carrier screening through Fulgent for 176 genes (reported on April 1, 2024) and was found to be a carrier of 1 condition.

Jane has not completed expanded carrier screening.

Discussion Regarding Reproductive Risks Based on Carrier Screening

I reviewed autosomal recessive inheritance and briefly reviewed each disease for which one gamete source was found to be a carrier. Detailed descriptions of each disease for which the egg source or sperm donor was found to be a carrier can be found on the carrier screening report. I reviewed the following risks:



1. There is a 50% chance that each genetic variant identified in the egg source or sperm source will be passed along to each future pregnancy.



2. If both the egg source and the sperm source are carriers of the same disease, there is a 25% risk that each child will inherit both variants and be at risk for the disease.



3. If one gamete source is a carrier of an autosomal recessive disease and the other gamete source is negative for that disease, the risk to have a child affected with that disease is significantly reduced, but a small residual risk remains.

Carrier Sperm Donor

		A	a
Carrier Egg Source	A	AA	Aa
	a	Aa	aa

A: Functional gene
a: Gene with variant

If both gamete sources are carriers of the same autosomal recessive disease, there is a:

- AA** 25% chance each child inherits both variant and may develop the disease
- Aa** 50% chance each child is a carrier
- aa** 25% chance each child is a non-carrier and unaffected

Carrier Screening Results

Donor BFA0184 has a positive result for the condition(s) listed below. Jane has not been tested for the condition(s), and reproductive risks depend on population and/or ethnicity- based carrier frequencies.

Disease (Gene)

Congenital disorder of glycosylation type 1a (PMM2)

X-Linked Inheritance

Carrier screening of the egg source for X-linked conditions may be considered. For most X-linked genetic conditions, only the egg source needs to be a carrier for there to be a risk of having an affected child. If the egg source is identified as a carrier, offspring with XY chromosomes (assigned male at birth) would have a 50% chance of being affected, and offspring with XX chromosomes (assigned female at birth) would have a 50% chance of being carriers.

Limitations of Carrier Screening

1. The detection rate for genes included on carrier screening panels is not 100%. A negative carrier screen reduces the chance of having an affected child but does not eliminate the chance.
2. Carrier screening includes a limited number of inherited conditions. There are thousands of genetic conditions that are not included on carrier screening panels.
3. There is a 3-6% chance for a birth defect in any child, regardless of carrier screening results.
4. The discussion today was based upon the interpretation(s) provided by the carrier screening laboratory/ies, which may change as genetic and medical research evolves.

Summary

Today's consultation provided a summary of Donor BFA0184's carrier screening results. The decision about which donor to use and what type of fertility treatment to proceed with is that of the patient and physician.

Jane is aware that they may initiate carrier screening through Guided Genetics, our partner genetic counseling service in the UK, by emailing sara@guidedgenetics.com.

On behalf of Seattle Sperm Bank, I wish you the best on your family building goals. Please reach out to me with any questions.

Sincerely,



Brynn Persky, MS, CGC
Certified Genetic Counselor

Seattle Sperm Bank

(206) 588-1484 #610

genetics@seattlespermbank.com

*The counseling provided to the benefits and limitations of the genetic testing that the donor(s) completed. A comprehensive personal and family medical history and genetic risk assessment was not performed for the client. Your patient may be referred to a medical geneticist or genetic counselor for concerns related to their own health or family history.