February 6, 2025

Seattle Sperm Bank

MEDICAL UPDATE LETTER FOR DONOR #10625 (ARNIE) - 2025-02-06

Donor #10625 (Arnie) is a carrier for POLG-related disorders.

To Whom it May Concern,

We are writing to provide a medical update regarding Donor #10625 (Arnie).

Donor #10625 (Arnie) recently completed updated genetic carrier screening on behalf of a customer. Below you will find a summary of all relevant information.

Genetic Results:
Donor #10625 (Arnie) was identified as a carrier for a variant in the *POLG* gene, associated with POLG-related disorders. POLG-related disorders typically follow autosomal recessive inheritance. This means that to have a child affected with this condition, both the egg and sperm source must carry a variant in the POLG gene and pass the variants on to the child. However, in a minority of cases, heterozygous carriers of *POLG* variants have a condition called autosomal dominant progressive external ophthalmoplegia (PEO). PEO is characterized by weakness of the eye muscles and eyelids. Some people can also have muscle weakness in other parts of the body including the heart. They may also experience a lack of muscle coordination, which can lead to clumsy movements and difficulty with balance and walking. Features typically present in early to mid-adulthood. The clinical features can vary widely even within the same family.

Please see the attached result report for additional details.

We contacted the donor to review his POLG carrier result and to obtain an updated medical history. The donor denied a personal or family history of PEO and of clinical features related to PEO.

Interpretation:

- All children/embryos resulting from this donor have a 50% chance to inherit the donor's variant.
- A minority of individuals who inherit this variant are at risk of autosomal dominant progressive external ophthalmoplegia.
- If the egg source of any child/embryo is also a carrier of a variant in the *POLG* gene, there is a 25% chance that a child/embryo will inherit both variants and be affected with POLG-related disorders (see the result report for details regarding these disorders).

Next Steps for Families who have Utilized the Donor:
Please share this letter and the attached result report with your fertility care provider and/or your child's pediatrician, who can suggest appropriate referrals when warranted. Screening of the egg source for *POLG* variants would provide more specific information about the chance to have a child/embryo affected with autosomal recessive POLG-related disorders. If the egg source has been screened for *POLG* and results are negative, the risk to have a child/embryo affected with autosomal recessive POLG-related disorders is substantially reduced.

Do not hesitate to reach out if questions arise or if clarification would be helpful. You are welcome to schedule a phone consultation with one of Seattle Sperm Bank's certified genetic counselors if you would like to discuss this information over the phone.

In partnership,

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