August 26, 2026

Seattle Sperm Bank

MEDICAL UPDATE LETTER FOR DONOR #20055 (CIARAN) - 2026-08-26

Donor 20055 (Ciaran) was recently reported to be a carrier of a genetic condition, called biotinidase deficiency. This condition was not included on his prior carrier screening. This letter is intended for all recipients and facilities who have utilized vials from Donor 20055 (Ciaran) to make them aware and provide follow-up suggestions.

To Whom it May Concern,

We are writing to provide a medical update regarding Donor #20055 (Ciaran).

Donor 20055 (Ciaran) was determined to be a carrier of a variant in the BTD gene. Specifically, he is heterozygous (one copy) for common variant c.1270G>C (p.Asp424His). In the literature, this variant is often noted as D444H, and is associated with partial biotinidase deficiency.

The BTD gene is associated with autosomal recessive biotinidase deficiency. Affected individuals inherit two variants in the BTD gene, one from the egg source and one from the sperm source. Carriers of BTD, like Donor 20055 (Ciaran), are not expected to exhibit symptoms.

With biotinidase deficiency the body has difficulty recycling an essential B vitamin called biotin. The symptoms of biotinidase deficiency are variable. In **profound biotinidase deficiency**, symptoms typically present during the first few months of life. If left untreated, affected individuals develop low muscle tone (hypotonia), skin rashes, hair loss (alopecia), seizures, hearing and vision loss, difficulty with balance and coordination (ataxia), and developmental delay. **Partial biotinidase deficiency** is a milder form of the condition. Affected individuals typically do not have any signs or symptoms of the condition (asymptomatic). However, if untreated, symptoms of partial BTD may appear during times of illness or stress. These symptoms may include hypotonia, skin rash, and alopecia. Partial biotinidase deficiency is caused by the combination of the mild D444H variant and a second, more severe disease-causing variant in the BTD gene. Of note, when an individual inherits two copies of the D444H variant (homozygous) from the sperm source and the egg source, they have mildly reduced enzyme levels but are not expected to have clinical symptoms of full or partial biotinidase deficiency.

Interpretation:
All children/embryos resulting from Donor 20055 (Ciaran) have a 50% chance to have inherited the BTD c.1270G>C variant. Children/embryos who inherit one copy of the variant would be asymptomatic carriers, just like donor.

Next Steps for Families who have Utilized the Donor:
Please share this letter and the attached report with your doctors. The egg source of any embryos/children resulting from Donor 20055 (Ciaran) may consider carrier screening for variants in the BTD gene.

- If the egg source of any child/embryo is a carrier of a variant in the BTD gene, there is a 1 in 4 (25%) chance that each resulting child/embryo will inherit *both* variants and may be affected with partial biotinidase deficiency.
    - The reproductive risk and clinical presentation may be further refined based on the BTD variant identified in the egg source.
- If the egg source of any child/embryo screens negative for mutations in the BTD gene, the risk to have a child affected with biotinidase deficiency is significantly 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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