

Donor	Bank	Jaar Rapid Alert	Aantal gezinnen	Informatie
Donor 1	ESB	2022	7	<p><u>First blockage:</u> Donor is a carrier of classic galactosemia. His gametes are permanently blocked 23MAY2022.</p> <p><u>Second blockage:</u> The donor is carrier of Shwachman-Diamond syndrome. His gametes were already permanently blocked in May 2022 due to his carrier status of classic galactosemia.</p>
Donor 2	ESB	2022	19	A pathogenic variant in the PHA gene has been identified in donor and he is a carrier of PKU why his gametes are permanently blocked 11JUL2022.
Donor 3	ESB	2022	10	Donor is a carrier of autosomal recessive inherited mild-to-moderate sensorineural hearing loss due to a STRC deletion and according to the current interpretation of the Danish legislation his gametes are permanently blocked 02SEP2022.
Donor 4	ESB	2022	14	The donor is a carrier of congenital disorder of glycosylation type Ia. His gametes are permanently blocked. 15DEC2022
Donor 5	ESB	2023	17	Hip dysplasia in a donor confers an increased risk to his offspring. His gametes are permanently blocked 18JAN2023.
Donor 6	ESB	2023	9	Donor is a carrier of c.10955delC(p.Pro3652Glnfs*2) in the PKHD1 gene. His gametes are to be permanently blocked 20MAR2023.
Donor 7	ESB	2023	11	A deletion of exon 11-14 in the GLI2 gene in a donor child does confer an increased risk to donor's offspring. His gametes are permanently blocked 24MAR2023.

Donor 8	ESB	2023	8	Donor is compound heterozygous for hemochromatosis. His gametes are therefore permanently blocked 15JUN2023.
Donor 9	ESB	2023	8	A duplication of 22q11 in the donor might confer an increased risk to his offspring. His gametes are permanently blocked 04JUL2023.
Donor 10	ESB	2023	12	The genetic examination of the donor shows that he has a deletion in exon 16-29 of the STRC gene. Hearing impairment in a donor child may confer an increased risk to donor's offspring. Donor's gametes are blocked 05JUL2023.
Donor 11	ESB	2023	11	Donor investigation shows donor to be a carrier of 57kb deletion in the CTNS-gene. A 57 kb deletion in CTNS-gene in a donor child does confer an increased risk to donor's offspring. Donor's gametes are to be permanently blocked 08AUG2023
Donor 12	ESB	2023	38	TP53 variant in a donor does confer an increased risk to donor's offspring. His gametes are permanently blocked 30OCT2023.
Donor 13	ESB	2023	12	The donor has been under investigation and the results have shown that he is a carrier of neonatal Zellweger syndrome, since he is heterozygous for the mutation in the PEX6 gene. Zellweger syndrome in a donor child does confer an increased risk to donor's offspring. His gametes are permanently blocked 07DEC2023.
Donor 14	ESB	2023	11	Genetic testing showed that Donor is carrier of one of the two RAG2 variants. RAG2 deficiency results in the severe phenotype Severe combined immune deficiency SCID. Due to the fact that donor is known carrier of a recessive gene variant he is permanently blocked 08NOV2023.

Donor 15	ESB	2024	7	Isovaleric acidemia in a donor child, and subsequent genetic analysis showing that donor is heterozygous carrier of a pathogenic variant in the IVD gene; c.158G>A, p.Arg53His, does confer an increased risk to donor's offspring. His gametes are therefore permanently blocked 20DEC2023.
Donor 16	ESB	2024	10	The donors Family history reveals that the PD has an unspecified hernia, his identical twin brother was born with coarctation of his aorta and his paternal aunt was born with only one kidney. Hydronephrosis in a donor child does confer an increased risk to donor's offspring. His gametes are therefore permanently blocked 31Jan2024.
Donor 17	ESB	2024	18	MSH2 variant in a donor does confer an increased risk to donor's offspring. His gametes are to be permanently blocked.
Donor 18	ESB	2024	14	The pathogenic variant in the GAA gene was identified in the donor. Biallelic pathogenic variants in the GAA gene cause Pompe disease. A pathogenic variant in the GAA gene does confer an increased risk to a donor child. His gametes are to be permanently blocked.
Donor 19	ESB	2024	8	Pathogenic MYBPC3 variant in a fetus and found in the donor confers an increased risk to donor's offspring. His gametes are permanently blocked 07NOV2024.
Donor 20	ESB	2024	17	Cardiomyopathy in a donor child and subsequent diagnosis of heterozygosity of a pathogenic variant in APLK3 gene in the donor confers an increased risk to donor's offspring. The donor is blocked and his gametes can no longer be used. The case is closed and reported to the Danish Patient Safety Authorities. 19NOV2024

Donor 21	ESB	2025	17	Investigation of donor shows a 109kb deletion involving the NRXN1 gene.Donor will be permanently blocked.21JAN2025
Donor 22	Cryos	2025	9	Donor has been tested and carries a variant in USH2A. The risk of an affected child is lower than 1%. Donor gametes were blocked for new customers (<1% recurrence risk) 02NOV2022 but can be used for siblings. Reported to the Danish Patient Safety Authority 25FEB2025.
Donor 23	ESB	2025	27	The donor has been investigated and has an extraordinary genetic constellation since he has 3 copies of the FBN1 gene: 2 normal FBN1 genes and a copy of FBN1 with deletion of exon 1-4. Donor gametes are blocked 30APR 2025. We recommend that donor conceived children of donor are referred for paediatric evaluation to assess the relevance of genetic counselling and examination.
Donor 24	ESB	2025	10	We can conclude that the donor has a genotype with a deletion of SMN1 on one chromosome and two copies of SMN1 on the other chromosome.The donor will be permanently blocked 20JAN2025.
Donor 25	ESB	2025	19	The donor is found to be a healthy carrier of a pathogenic variant in the CYP21A2 gene. Non-classical congenital adrenal hyperplasia (CAH) due to compound heterozygous mutations in the CYP21A2 gene in a donor child confers an increased risk to donor's offspring. His gametes are permanently blocked. The case is reported to the Danish Patient Safety Authorities. 07JUL2025.
Donor 26	ESB	2025	9	Deletion of the CYP21A2 gene in a donor does confer an increased risk to donor's offspring. His gametes are to be permanently blocked. The case is closed and reported to the Danish Patient Safety Authorities. 19AUG2025

Donor 27	ESB	2025	12	A pathogenic (class V) genetic variant was observed in the Thyroglobulin (TG) gene. Congenital hypothyroidism in a donor child does confer an increased risk to donor's offspring. His gametes are to be permanently blocked. The case is closed and is to be reported to the Danish Patient Safety Authorities. 13OCT2025.
Donor 28	ESB	2025	22	The donor is a carrier of Metachromatic leukodystrophy (MLD), since a pathogenic genetic variant in the Arylsulfatase A (ARSA) gene is identified in a heterozygous state. A pathogenic genetic variant :NM_000487.6:c.917C>T, identified in the donor does confer an increased reproductive risk. His gametes are to be permanently blocked. 20OCT2025.
Donor 29	ESB	2025	15	The donor has been under investigation and the results have shown that he is a healthy carrier of Spinal Muscular Atrophy, since MLPA analysis shows a deletion of one of the SMN1 genes.

Bron: FAGG