

## Supplementary material:

**Table 1:** Summary of the PGD data for monogenic diseases.

Indications	DMD	DM1	OTCD	FCMD	ALD	Leigh syndrome	Total
Cycles to OR	59	31	6	1	1	1	99
Number infertile	24	9	1	1	1	1	37
Female age (average)	38	36	42	38	41	39	38
Cancelled before IVF/ICSI	1	1	0	0	0	0	2
ART method							
IVF	0	0	0	0	0	0	0
ICSI	58	28	6	1	1	1	95
IVF+ICSI	0	0	0	0	0	0	0
Unknown	0	2	0	0	0	0	2
Cancelled after IVF/ICSI	1	0	1	0	0	0	2
Cycles to PGD	57	30	5	1	1	1	95
Zona breaching							
AT drilling	0	0	0	0	0	0	0
Laser drilling	57	23	0	1	1	1	83
Mechanical	0	0	0	0	0	0	0
Laser drilling + Mechanical	0	0	5	0	0	0	5
Unknown	0	0	0	0	0	0	0
Biopsy method							
Polar body biopsy	0	0	0	0	0	0	0
Cleavage aspiration	57	23	5	1	1	1	88
Cleavage extrusion	0	0	0	0	0	0	0
Blastocyst	0	0	0	0	0	0	0
Indications	DMD	DM1	OTCD	FCMD	ALD	Leigh syndrome	Total
Embryology							
COCs (mean / OR)	614 (10.4)	360 (11.6)	52 (8.7)	8 (8.0)	13 (13.0)	3 (3.0)	1050 (10.6)
Fertilized	395	224	29	5	7	1	661
Biopsied	331	186	25	4	7	1	554
Frozen before biopsy	0	0	0	0	0	0	0
Successfully biopsied	330	174	25	4	7	1	541
Diagnosed (mean / OR)	254	101	22	-	5	1	383
Transferable (mean / OR)	175	48	12	-	5	0	240
Transferable rate (% / diagnosed)	68.9	47.5	54.5	-	100	0	62.7
Transferred	125	30	7	-	5	0	167
Clinical outcome							
Cycles to ET (% / OR)	90 (153.0)	19 (61.3)	5 (83.3)	0 (0.0)	3 (300.0)	0 (0.0)	117.0 (118.2)
hCG positive	14	3	0	0	2	0	19
Positive FHB	12	1	0	0	1	0	14
Number of FHB	13	1	0	0	1	0	15
Clinical pregnancy rate (% / OR)	20.3	3.2	0	0	100	0	14.1
Clinical pregnancy rate (% / ET)	13.3	5.3	0	0	33.3	0	12.0
Implantation rate (FHB / embryos transferred)	10.4	3.3	0	0	20	0	9.0
Deliveries	9	0	0	0	1	0	10
Miscarriages	5	2	0	0	1	0	8
Ectopic pregnancies	0	1	0	0	0	0	1
Twins	1	0	0	0	0	0	1

DMD, Duchenne muscular dystrophy;DM1, myotonic dystrophy type 1;OTCD, Ornithine transcarbamylase deficiency; FCMD, Fukuyama congenital muscular dystrophy;ALD, Adrenoleukodystrophy; OR, oocyte retrieval; AT, acid Tyrode's solution; COC, cumulus-oocyte complex; ET, embryo transfer; ART, assisted reproduction technology; FHB, fetal heart beat.

**Table 2:** Summary of the PGD data for chromosomal abnormalities.

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Indications	Rob, male carrier	Rob, female carrier	Rcp, male carrier	Rcp, female carrier	Total
Cycles to OR	13	48	179	332	572
Number infertile	4	16	67	87	174
Female age (average)	43	39	39	39	40
Cancelled before IVF/ICSI	0	2	8	14	24
ART method					
IVF	0	11	61	76	148
ICSI	13	37	105	239	394
IVF+ICSI	0	0	5	3	8
Unknown	0	0	0	0	0
Cancelled after IVF/ICSI	1	5	11	20	37
Cycles to PGD	12	41	160	298	511
Zona breaching					
AT drilling	0	1	5	13	19
Laser drilling	10	20	61	156	247
Mechanical	2	20	94	129	245
Unknown	0	0	0	0	0
Biopsy method					
Polar body biopsy	0	0	0	3	3
Cleavage aspiration	10	38	130	262	440
Cleavage extrusion	2	3	30	33	68
Blastocyst	0	0	0	0	0
Indications	Rob, male carrier	Rob, female carrier	Rcp, male carrier	Rcp, female carrier	Total
Embryology					
COCs (mean / OR)	63 (4.8)	350 (7.3)	1322 (7.4)	2405 (7.2)	4140 (7.2)
Fertilized	34	225	922	1563	2744
Biopsied	11	166	636	1009	1822
Frozen before biopsy	13	0	80	228	321
Successfully biopsied	11	166	635	1007	1819
Diagnosed (mean / OR)	11	148	626	925	1710
Transferable (mean / OR)	3	42	177	214	436
Transferable rate (% / OR)	27.3	28.4	28.3	23.1	25.5
diagnosed)					
Transferred	2	23	114	153	292
Clinical outcome					
Cycles to ET (% / OR)	2 (15.4)	16 (33.3)	83 (46.4)	115 (34.6)	216 (37.8)
hCG positive	0	5	45	51	101
Positive FHB	0	4	40	33	77
Number of FHB	0	5	41	34	80
Clinical pregnancy rate (% / OR)	0	8.3	22.3	9.9	13.5
Clinical pregnancy rate (% / ET)	0	25	48.2	28.7	35.6
Implantation rate (FHB / OR)	0	21.7	36.0	22.2	27.4
embryos transferred)					
Deliveries	0	3	28	29	60
Miscarriages	0	2	12	22	36
Ectopic pregnancies	0	0	0	2	2
Twins	0	1	1	1	3

Rob, Robertsonian translocation; Rcp, Reciprocal translocation; OR, oocyte retrieval; AT, acid Tyrode's solution; COC, cumulus-oocyte complex; ET, embryo transfer; ART, assisted reproduction technology; FHB, fetal heart beat.

**Table 3:** Prenatal and postnatal diagnosis.

Methods	Total		Results		
			Normal	Abnormal	Failed
Prenatal diagnosis					
Amniocentesis	26	26	0	0	0
CVS	0	0	0	0	0
Cordocentesis	1	1	0	0	0
Unexamined	30				
Unknown	13				
Postnatal diagnosis					
Karyotype	3	3	0	0	0
DNA test	1	1	0	0	0

CVS, chorionic villus sampling.

**Table 4:** Method of delivery, gestational age, and infants born.

	Singletons	Twins	Total
No. deliveries	65	5	70
Method of delivery			
Vaginal	32	0	32
Caesarian	26	5	31
Unknown	7	0	7
Term at delivery			
Preterm	6	4	10
Term	57	1	58
Post term	0	0	0
Unknown	2	0	2
Sex			
Male	30	2	32
Female	32	8	40
Unknown	3	0	3
Mean birthweight (g)	2964	2356	2919