

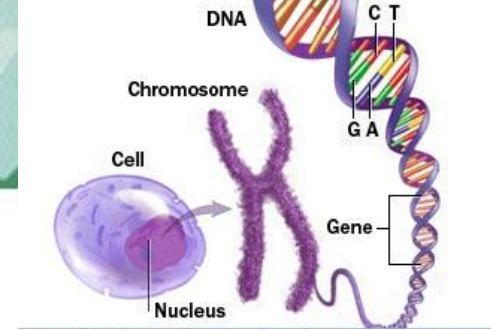
Obstetrical Outcome among Pregnancies Conceived after Preimplantation Genetic Diagnosis (PGD)

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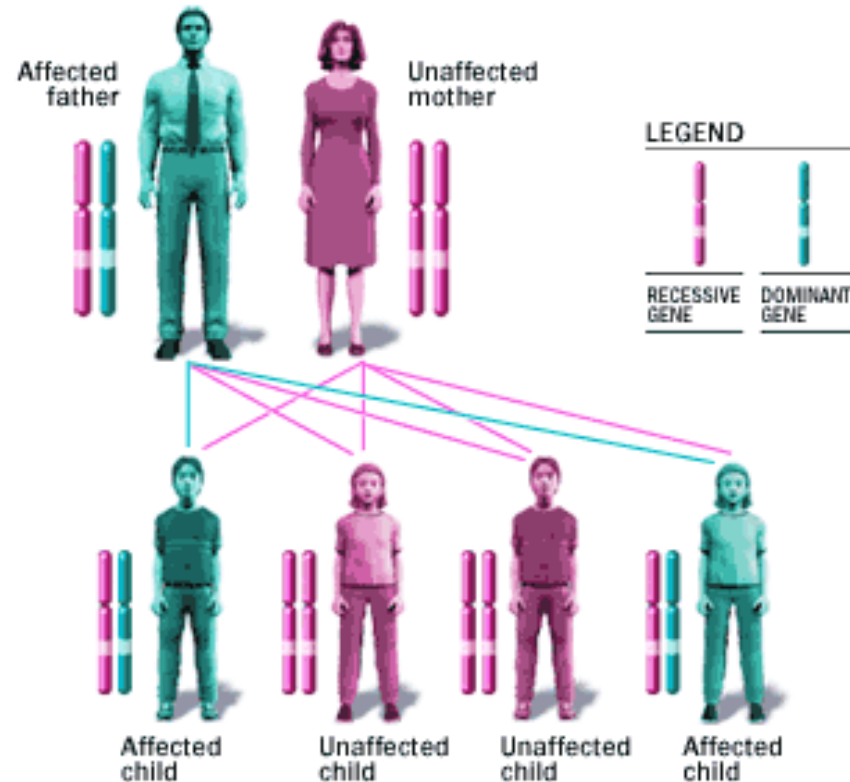


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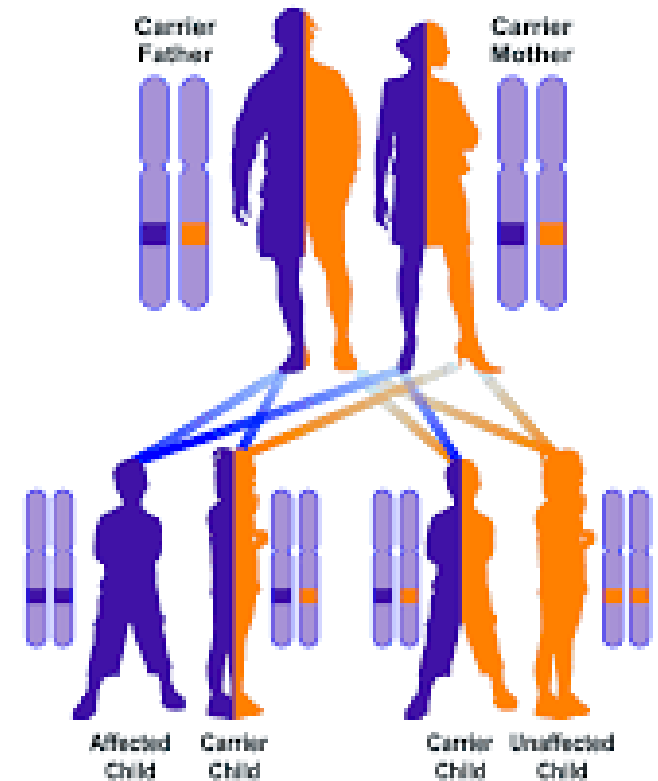
Genetic disorders



Autosomal dominant inheritance pattern



Autosomal Recessive



Pre-implantation genetic diagnosis (PGD)

PGD allows patients who are carriers of single-gene disorders or carriers of structural chromosome abnormalities (balanced translocations) to select unaffected embryos for transfer, and reduce the transmission of genetic disorders to the offspring.

Pre-implantation genetic diagnosis

- **PGD is considered in a similar fashion to prenatal diagnosis.**
- **PGD refers to the genetic profiling of embryos prior to implantation.**
- **PGD is an adjunct to assisted reproductive technology, and requires in vitro fertilization (IVF) to obtain oocytes or embryos for evaluation.**

IVF cycle



OPU

ET

gonadotropin administration

gonadotropin administration

administration



pre-

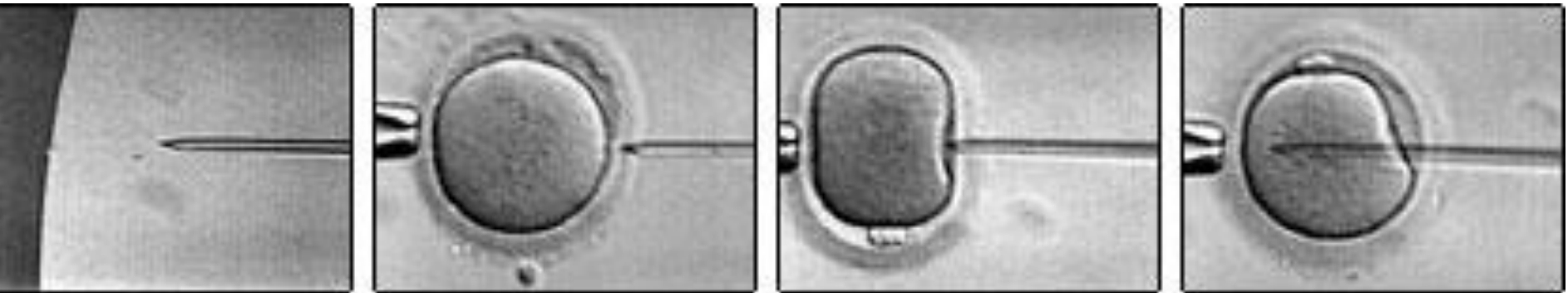
treatment cycle

Fertilization

IVF



ICSI

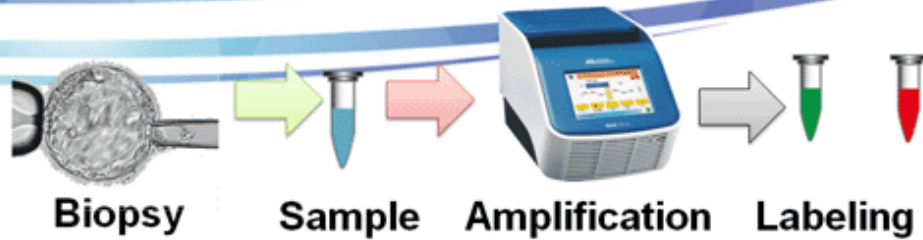


A single sperm being picked up and injected into a mature egg.

IVE



PGD-aCGH Workflow



Pregnancy outcome

□56 Obstetrical outcome after preimplantation genetics

Ginsberg N, Tur-Kaspa I, Cieslak-Janzen J, Rechitsky S, Pauling D, Horwitz A, Kuliev A, Verlinsky Y
Reproductive Genetic Institute, Chicago, IL, USA

Abstracts - 6th International Symposium on Preimplantation Genetics 2005

Conclusions: The obstetrical outcome after PGD is similar to that of other IVF pregnancies. PGD significantly reduced the incidence of multiple gestations.

995 children

No differences regarding mean term, prematurity (term <32 w and <37 w), mean birthweight, very low birthweight (<1500 g), perinatal death, major malformations and neonatal hospitalizations in singletons and multiples born following PGD versus ICSI were observed.

Neonatal follow-up of 995 consecutively born children after embryo biopsy for PGD
2012

**S. Desmyttere^{1,*}, M. De Rycke¹, C. Staessen¹, I. Liebaers¹,
F. De Schrijver¹, W. Verpoest², P. Haentjens³, and Maryse Bonduelle¹**

Neonatal outcome after preimplantation genetic diagnosis

Fertil Steril® 2014

Talia Eldar-Geva, M.D., Ph.D.,^{a,b} Naama Srebnik, M.D.,^{a,b} Gheona Altarescu, M.D.,^{b,c} Irit Varshaver, M.Sc.,^a Baruch Brooks, Ph.D.,^a Ephrat Levy-Lahad, M.D.,^{b,c} Ruben Bromiker, M.D.,^{b,d} and Michael S. Schimmel, M.D.^{b,d}

242 children born after PGD, 242 after ICSI and 733 after a spontaneous conception (SC).

Birth weight, pregnancy duration, and intrauterine growth for singletons.				
	PGD	ICSI	SC	<i>P</i> value ^a
Birth weight (g)	3,238 ± 514	3,062 ± 573	3,204 ± 504	.005
<2,500 (%)	7 (4.4)	19 (12)	27 (5.5)	.01
<1,500 (%)	1 (0.6)	2 (1.3)	7 (1.4)	.7
Pregnancy duration (wk)	38.8 ± 1.8	38.4 ± 2.1	39.2 ± 1.9	.005
<37 (%)	11 (7)	18 (11.4)	28 (5.7)	.05
<34 (%)	2 (1.3)	5 (3.2)	10 (2.0)	.5
<32 (%)	1 (0.6)	2 (1.3)	8 (1.6)	.6
<28 (%)	1 (0.6)	0	1 (0.2)	.5
Intrauterine growth				.001
IUGR (<10th percentile)	8 (5.1)	15 (9.5)	27 (5.5)	
Normal weight (10–90th percentiles)	124 (78.4)	125 (79.1)	421 (85.7)	
LGA (>90th percentile)	18 (16.5)	26 (11.4)	43 (8.8)	

Pre-term birth and low birth weight following preimplantation genetic diagnosis: analysis of 88 010 singleton live births following PGD and IVF cycles

2017

Sesh Kamal Sunkara^{1,*}, Belavendra Antonisamy², Hepsy Y. Selliah², and Mohan S. Kamath²

Table II PTB and LBW outcomes following PGD versus autologous IVF.

Outcome	n (%)		OR (95% CI)	aOR [†] (95% CI)
	PGD n = 439	Autologous IVF n = 87 571		
PTB (<37 weeks)	28 (6.4)	7968 (9.1)	*0.68 (0.46–0.99)	*0.66 (0.45–0.98)
Early PTB (<32 weeks)	4 (0.9)	1512 (1.7)	0.52 (0.20–1.40)	0.48 (0.18–1.31)
LBW (<2500 g)	24 (5.5)	8138 (9.3)	*0.56 (0.37–0.85)	*0.58 (0.38–0.88)
Very LBW (<1500 g)	4 (0.9)	1580 (1.8)	0.50 (0.19–1.34)	0.50 (0.18–1.35)

2016

Preimplantation genetic diagnosis: a national multicenter obstetric and

Obstetrical and neonatal outcomes following preimplantation genetic diagnosis (PGD), in vitro fertilization/intracytoplasmic sperm injection (IVF/ICSI), and spontaneous conception (SC) with or without existing monogenic disorders in the parents, Denmark 1999-2013.

Outcome	PGD ^a vs. IVF/ICSI ^b	PGD monogenic ^c vs. ICSI ^d	PGD monogenic ^c vs. SC ^e	PGD aberration ^f Vs. IVF ^g	PGD aberration ^f vs SC ^e	PGD monogenic ^c vs. SC monogenic ^h
Obstetrical						
Preeclampsia, odds ratio (95% CI)	1.2 (0.6; 2.4)	1.3 (0.5; 3.2)	1.2 (0.5; 3.2)	1.1 (0.4; 3.2)	1.2 (0.4; 3.3)	0.9 (0.4; 2.3)
Preterm primary rupture of membranes, odds ratio (95% CI)	1.3 (0.7; 2.4)	2.0 (0.9; 4.2)	2.1 (1.0; 4.4) ⁱ	0.6 (0.2; 2.0)	0.6 (0.2; 2.1)	1.4 (0.7; 3.1)
Placenta previa, odds ratio (95% CI)	2.1 (0.8; 5.8)	2.4 (0.6; 9.8)	9.4 (2.3; 38.7) ⁱ	1.8 (0.4; 7.4)	8.4 (2.0; 34.6) ⁱ	4.2 (1.1; 15.8) ⁱ
Abruption of placenta, odds ratio (95% CI)	1.2 (0.2; 8.3)	2.4 (0.3; 17.8)	3.6 (0.5; 26.4)	na	na	2.2 (0.2; 21.9)
Cesarean section, odds ratio (95% CI)	1.7 (1.1; 2.5) ⁱ	1.9 (1.1; 3.4) ⁱ	2.1 (1.1; 3.7) ⁱ	1.6 (0.9; 2.7)	2.0 (1.1; 3.5) ⁱ	1.4 (0.9; 2.2)
Induction of labor, odds ratio (95% CI)	0.9 (0.6; 1.5)	1.0 (0.5; 2.0)	1.2 (0.6; 2.2)	0.8 (0.4; 1.7)	1.0 (0.5; 2.0)	1.1 (0.7; 1.9)
Neonatal						
Gestational age (d), mean difference (95% CI)	-1.2 (-4.2; 1.8)	-5.1 (-9.1; -1.1) ⁱ	-6.3 (-9.5; -3.2) ⁱ	3.2 (-1.25; 7.7)	0.0 (-3.3; 3.3)	0.2 (-3.4; 3.8)
Preterm birth (<37 wk), odds ratio (95% CI)	1.3 (0.8; 2.1)	1.7 (0.9; 3.2)	1.8 (0.9; 3.5)	0.9 (0.4; 1.9)	1.2 (0.5; 2.8)	1.2 (0.6; 2.4)
Birth weight (g), mean difference (95% CI)	-10 (-115; 96)	-149 (-293; 5.4)	-178 (-311; -45) ⁱ	155 (1; 310) ⁱ	50 (-88; 189)	20 (-111; 151)
Low birth weight (<2500 g), odds ratio (95% CI)	1.2 (0.7; 2.0)	2.0 (1.1; 3.9) ⁱ	2.3 (1.1; 4.5) ⁱ	0.4 (0.2; 1.2)	0.5 (0.2; 1.5)	0.8 (0.4; 1.8)
Length at birth (cm), mean difference (95% CI)	-0.2 (-0.8; 0.4)	-0.9 (-1.8; -0.1)	-1.1 (-1.7; -0.4) ⁱ	0.6 (-0.2; 1.5)	0.3 (-0.4; 0.9)	0.6 (-0.2; 1.5)
Apgar score: below 7 (%), odds ratio (95% CI)	1.1 (0.3; 4.3)	1.9 (0.5; 8.0)	1.8 (0.5; 8.0)	na	na	0.5 (0.1; 3.3)
Neonatal admission, odds ratio (95% CI)	0.9 (0.5; 1.5)	1.4 (0.8; 2.6)	1.4 (0.8; 2.7)	0.4 (0.2; 1.1)	0.5 (0.2; 1.3)	1.2 (0.2; 2.4)
Length of neonatal admission (d), mean difference (95% CI)	20 (13; 27) ⁱ	30 (22; 39) ⁱ	31 (24; 38) ⁱ	-9 (-24; 5)	-7 (-19; 5)	13 (-11; 38)
Malformations, odds ratio (95% CI)	1.5 (0.9; 2.5)	1.3 (0.6; 2.8)	1.3 (0.6; 2.8)	1.7 (0.8; 3.6)	1.8 (0.8; 3.8)	1.2 (0.6; 2.2)
Stillbirth, odds ratio (95% CI)	na	na	na	na	na	na
Perinatal death, odds ratio (95% CI)	na	na	na	na	na	na
Infant death, odds ratio (95% CI)	na	na	na	na	na	na
Placenta weight (g), mean difference (95% CI)	13 (-21; 49)	-37 (-86; 12)	-17 (-54; 19)	68 (19; 119)	71 (33; 109) ⁱ	60 (25; 97) ⁱ



700 PGD pregnancies

**Oocyte- IVF/ICSI-Day3/4/5Bx-
FISH/PCR/CMA**

Oocyte/embryo quality

**Pregnancy (maternal and neonatal)
outcomes-Incomplete**



THANK YOU

Pre-implantation genetic diagnosis

Its main advantage is that it avoids selective pregnancy termination, as the method makes it highly likely that the baby will be free of the disease under consideration.