



### The role of the PGD Consortium

Joyce Harper  
Chair of the ESHRE PGD Consortium

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### Overview

- Aims
- Membership
- Data
- Pregnancies
- Guidelines
- Training courses/education/post congress workshops
- Working groups
- PGS task force
- Web site
- Steering committee



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### ESHRE PGD Consortium - Aims

- To survey the availability of PGD
- To collect prospectively and retrospectively data on the accuracy, reliability and effectiveness of PGD
- To initiate follow-up studies of pregnancies and children born
- To produce guidelines and recommended PGD protocols
- To formulate a consensus on the use of PGD
- To educate in the science of genetics and reproduction



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### Members by country I-X

Argentina:	1	Netherlands:	5
Australia:	2	Poland:	1
Brazil:	1	Portugal:	1
Belgium:	6	Russia:	1
Bulgaria:	1	Serbia:	1
Czech Republic:	2	Singapore:	1
Denmark:	3	South Africa:	1
Egypt:	1	Spain:	13
Germany:	9	Sweden:	2
Japan:	5	Thailand:	2
Finland:	2	Taiwan:	1
France:	3	Turkey:	3
Greece:	5	UK:	7
India:	2	Ukraine:	1
Israel:	4	United Arab Emirates:	1
Italy:	4	USA:	7
Korea:	2		
		TOTAL	101




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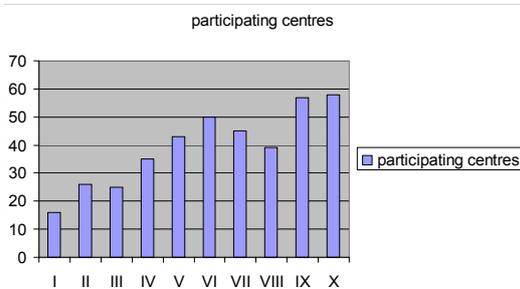
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### Survey the availability of PGD




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## Data Collection – 13 years

- ESHRE Preimplantation Genetic Diagnosis (PGD) Consortium (1999). Preliminary assessment of data from January 1997 to September 1998 ESHRE PGD Consortium Steering Committee. Human Reproduction, 14: 3138-3148.
- ESHRE PGD Consortium Steering Committee (2000) ESHRE Preimplantation Genetic Diagnosis (PGD) Consortium: data collection II (May 2000). Hum. Reprod. 15, 2673-2683.
- ESHRE PGD Consortium Steering Committee (2002) ESHRE Preimplantation Genetic Diagnosis (PGD) Consortium: data collection III (May 2001). Hum Reprod., 17, 233-246.
- Sermon, K., Moutou, C., Harper, J., Geraedts, J., Scriven, P., Wilton, L., Maqil, M.-C., Michiels A, Viville, S., De Die, C. (2005) ESHRE PGD Consortium data collection IV: May-December 2001, Human Reproduction, 20(1):19-34.
- Harper, J.C, Beelaert, K, Geraedts, J., Harton, G., Kearns, W.G, Moutou, C., Murljewerff, N., Repping, S., SenGupta, S., Scriven, P.N., Traeger-Synodinos, J., Vesela, K, Wilton, L., Sermon, K.D. (2006) ESHRE PGD Consortium data collection V: Cycles from January to December 2002 with pregnancy follow-up to October 2003, Human Reproduction, 21, 3-21
- Sermon, K.D, Michiels, A, Harton, G, Moutou, C, Repping, S, Scriven, P.N, SenGupta, S, Traeger-Synodinos, J, Vesela, K, Viville, S, Wilton, L, Harper, J.C (2007) ESHRE PGD Consortium data collection VI: Cycles from January to December 2003 with pregnancy follow-up to October 2004, Hum Reprod. 22(2):323-36
- Harper, J.C, De Die, C, Goossens, V, Harton, G, Moutou, C, Repping, S, Scriven, P., SenGupta, S., Traeger-Synodinos, J., Viville, S., Wilton, L., Sermon, K.D. (2007) ESHRE PGD Consortium data collection VII: Cycles from January to December 2004 with pregnancy follow-up to October 2005. Hum Reprod
- Goossens, V, Harton, G, Moutou, C, Scriven, P.N, Traeger-Synodinos, J, Sermon, K, Harper, J.C (2008) ESHRE PGD Consortium data collection VIII: Cycles from January to December 2005 with pregnancy follow-up to October 2006, Human Reproduction, 23(12):2629-45
- Goossens, V, Harton, G, Moutou, C, Traeger-Synodinos, J, Van Rij, M and Harper, J.C (2009) ESHRE PGD Consortium data collection IX: cycles from January to December 2006 with pregnancy follow-up to October 2007

www.eshre.com




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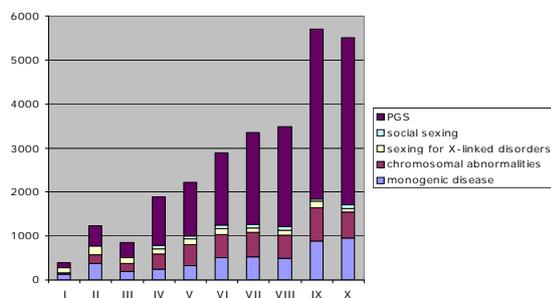
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## Evolution of cycle data




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### Cumulative data I-X

	Cycles to OR	No. embryos biopsied	No. embryos transferred	Embryo transfer procedures	Pregnancy rate (per OR and per ET)
Monogenics	4471	26490	6749	3527	23% per OR 29% per ET
Chromosomes	4128	26733	4637	2634	17% per OR 26% per ET
Sexing X linked	1141	7137	1555	855	19% per OR 26% per ET
Aneuploidy	16852	90611	21425	12008	19% per OR 26% per ET
Social sexing	671	4283	995	492	21% per OR 29% per ET



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### Top 10 monogenics

- Recessive
  - $\beta$ -thalassaemia
  - Cystic Fibrosis
  - Spinal muscular atrophy
  - Sickle cell disease
- Dominant
  - Huntington's disease
  - Myotonic dystrophy
  - Charcot-Marie-Tooth disease
- Sex linked (specific diagnosis)
  - Fragile X syndrome
  - Duchenne muscular dystrophy
  - Haemophilia



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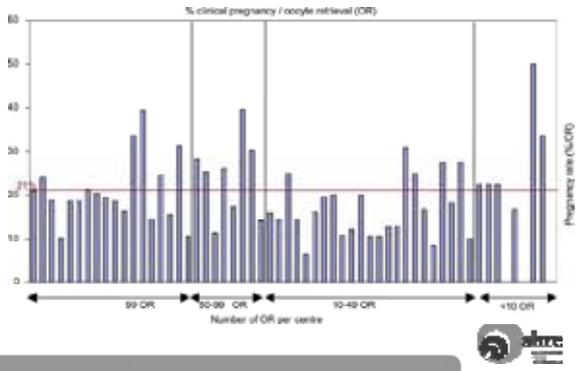
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### Pregnancy rate by centre Data IX




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### PGD Guidelines

- ESHRE guidelines on developing guidelines
- Assessment of previous version and decision to develop a new format
- Original document broken down into four new guidelines
  - Organization of a PGD center
  - PCR-based PGD with Transport PGD
  - FISH-based PGD with Transport PGD
  - Biopsy for PGD in conjunction with the Embryology SIG




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## Consortium/SIG training

- Biopsy and FISH workshop: Prague, 2006
- Basic genetics for ART practitioners:
  - Brussels, 2006
  - London, 2007
  - Athens, 2008
  - Strasbourg, 2009
- Biopsy trouble shooting day: Post congress course Lyon 2007
- Accreditation workshop: Brno, 23-24 October 2008
- **2009/2010/2011**
- Use of arrays in PGD – post congress course, 2<sup>nd</sup> July 2009
- Quality Management – two day course in London, 22/23 March 2010
- Celebration of 20 years of PGD – post congress course, 1<sup>st</sup> July 2010
- Basic genetics for ART practitioners – Portugal, 2010
- Biopsy post congress workshop 2011



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## Celebration of 20 years of PGD, 1<sup>st</sup> July 2010

- **Past, present and future**
- The history of PGD - Joyce Harper
- The early years: pre clinical PGD – Marilyn Monk
- The evolution of technologies over 20 years – Alan Handyside
- Embryo biopsy – Anna Veiga
- What is reality and what is not – Mark Hughes
- The value of the polar bodies – Arver Kuliev
- PGD for infertility – Santiago Munne
- PGD – from diagnosis to treatment - Cristina Magli
- **The Patients Perspective**
- Prenatal to PGD – which one to choose – Joe Leigh Simpson
- Follow up of the PGD babies – Alison Lashwood
- The patients perspective - Inge Liebaers
- A PGD success story (SMA) – A patient talk by the Galloway family (Gary Harton)
- PGD for HLA matching- how we cured our child following PGD treatment – Ponchirolli family (Francesco Fiorentino)
- **Ethics and the future**
- What PGD has taught us about preimplantation development? Leeanda Wilton
- PGD: Dynamics and Ethics - Guido de Wert
- Panel discussion – is GATTACA almost here?



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## Consortium working groups

- Aneuploidy screening: Chair Sjoerd Repping
  - CLOSED – ESHRE PGS task force
- Misdiagnosis: Chair Joanne Traeger-Synodinos
  - Paper on causes of misdiagnosis – Wilton et al, 2009
  - Study on follow up of untransferred embryos
- Diagnostic laboratory accreditation: Chair Katerina Vesela
  - Workshops 2008/2010
  - Beginners guide towards accreditation – Harper et al, 2010
  - Questionnaire
  - Accreditation advisory panel
- Database: Chair Celine Moutou
  - Setting up a new database
  - Questionnaire on freezing
- Guidelines: Chair Gary Harton
  - Four guidelines prepared
- Molecular methods: Chair Francesco Fiorentino
  - Database of methods



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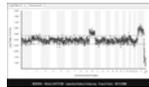
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## ESHRE PGS task force

- Set up in 2007
- ESHREs first involvement in a clinical trial
- Assess efficacy of PGS for advanced maternal age using polar body biopsy and analysis of 24 chromosomes (array)
- Two stages – pilot and randomised controlled trial
- Task force members
  - Paul Devroey, Luca Gianaroli, Alan Handyside, Sjoerd Repping, John Collins, Catherine Staessen, Katerina Vesela, Joyce Harper, Veerle Goossens, Joep Geraedts, Markus Montag and Andreas Schmutzler



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## Pilot study

- Feasibility study
  - Can analysis of both polar bodies be completed within a time period that allows for fresh transfer?
  - Ensure reliable identification of the chromosomal status of an oocyte in at least 90% of polar body biopsy attempts
  - Test the feasibility of a multicentre randomized trial based on the technology used in the pilot study
- Two centres with polar body biopsy and PGS experience
  - SIMMER, Bologna, Italy (Cristina Magli, Luca Gianaroli)
  - University of Bonn, Germany (Markus Montag, Hans van der Ven)
- Both polar bodies and untransferred zygotes analysed using 24sure with no age restriction
  - BlueGnome, Cambridge, UK
- Data analysed by third party
  - Sjoerd Repping, Academic Medical Centre, Amsterdam
- Started September 2009, Geraedts et al, 2009
- Present data ESHRE Rome 2010



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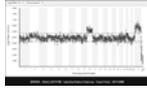
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### Randomised controlled trial

- If pilot shows technology is feasible –
- RCT
- Multi-centre
- At least six centres in different EU countries
- Advanced maternal age
- Polar body biopsy allows
  - Fresh transfer
  - Less invasive biopsy
  - Hopefully more reliable than cleavage or blastocyst biopsy (mosaicism)
- If trial shows that PGS improves delivery rates – will be major step in IVF treatment



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### Consortium – where we are today

- History of Consortium, Focus on Reproduction, Jan 2007
- Two types membership (Full and Associate)
- Newsletter to all staff of clinics who are members
- Paediatric follow up
- Training, education, communication
- Working groups
  - accreditation, misdiagnosis, database, guidelines, molecular
- EQA FISH and PCR
- Closed consortium annual meeting, 4-7pm, Saturday before ESHRE
  - Saturday 26<sup>th</sup> June, 4-7pm, 2010, Rome



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### Consortium web site

- PGD consortium members and the treatments they offer
- Summary of our activities
- Recommended reading
- Publications of the consortium
- Guidelines
- Newsletters
- Training and education
- Cumulative tables of results
- Statutes
- How to become a member
- [www.eshre.com/emc.asp?pageld=390](http://www.eshre.com/emc.asp?pageld=390)



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## Acknowledgement

- Thanks to
  - All clinics who send in data
  - Steering committee
- Invites
  - Email list – all staff at centre
  - Centres to join
  - New members to working groups



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## Steering Committee

1997



2010

Joyce Harper, UK, Chair  
Gary Harton, USA, Deputy  
Céline Moutou, FR  
Sioban SenGupta, UK  
Joanne Traeger-Synodinos, GR  
Katerina Vesela, CZ

2009



Tugce Pehlivan, SP  
Francesco Fiorentino, IT  
Edith Coonen, N  
Martine De Rycke, BE  
Pam Renwick, UK  
Veerle Goossens, BE



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