

Joint Research Centre (JRC)

Where is Preimplantation Genetic Diagnosis going in Europe?



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The European Commission's Joint Research Centre...

provides customer-driven **scientific and technical support** for the conception, development, implementation and monitoring of **EU policies**

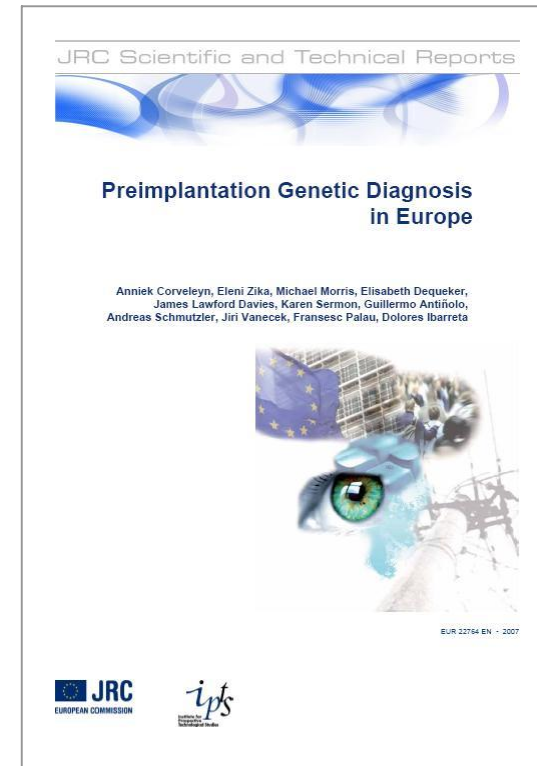
The JRC functions as a **reference centre of science and technology for the Union**

Close to the policy-making process, it serves the common interest of the Member States, while being **independent of special interests, whether private or national.**



Why the JRC is looking into this

- A comprehensive picture of PGD practice and provision in Europe was lacking
 - concerns over quality assurance for these services
 - lacking knowledge of impact of different regulatory frameworks – international flow



How the JRC looked into this

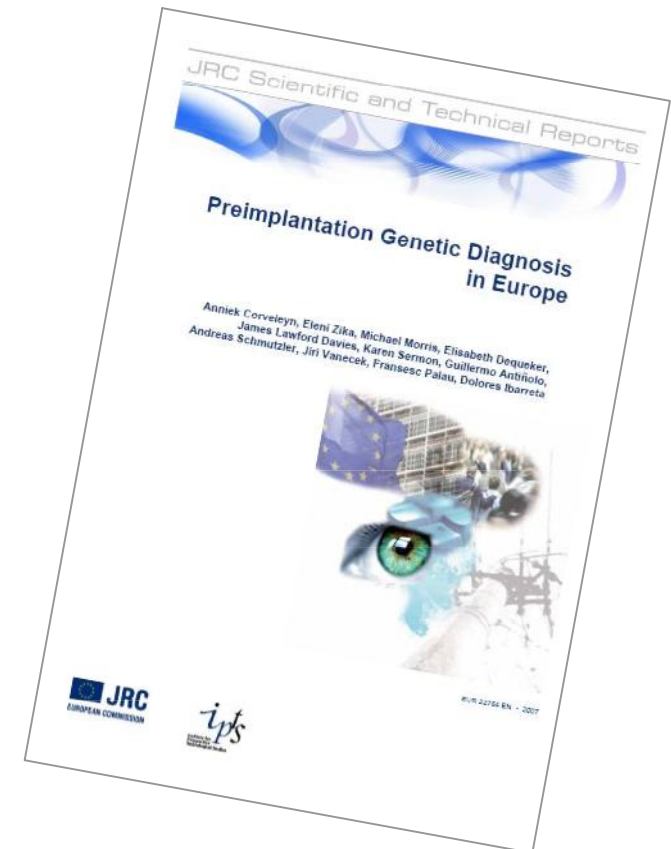
March 2005: workshop ESHRE/ESHG/JRC-IPTS

End of 2006: Study launched

December 2008: Publication of the report

Objectives of the “Preimplantation Diagnosis in Europe” study:

1. obtain a clear picture of current PGD practice in Europe;
2. carry out a comparative review of the different regulatory frameworks at Member State level



The study's methodology

SURVEY of centres and clinics potentially performing PGD or offering PGD-related services in Europe identified by ESHRE and from the EuroGentest quality assurance survey and expert consultation

ON-LINE QUESTIONNAIRE, which was sent to 169 those European PGD and IVF clinics identified

EXPERT INTERVIEWS provision in Belgium, the Czech Republic, France, Germany, Greece, Ireland, the Netherlands, the Slovak Republic, Spain, Switzerland and the UK.

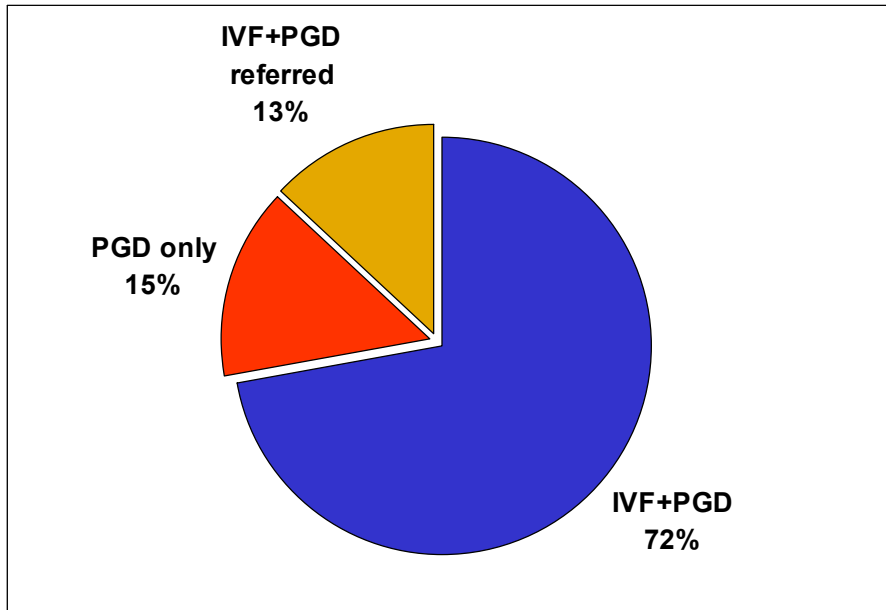


PROJECT TEAM

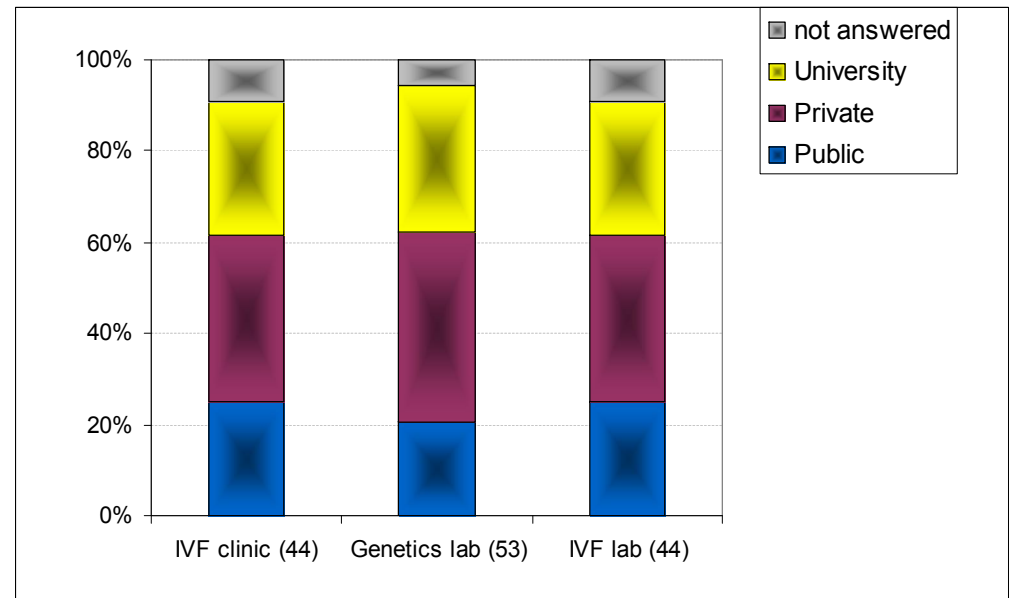
Elisabeth Dequeker, Anniek Corveleyn, Michael Morris, Nick Nagels
(EuroGentestNetwork of Excellence)
James Lawford Davies **(Epan – GIG-PET, UK)**
Guillermo Antiñolo **(Hospital Universitario Virgen del Rocio, Spain)**
Fransesc Palau **(Institut de Biomedicina de Valencia – CSIC, Spain)**
Andreas Schmutzler **(University of Kiel, Germany)**
Jiri Vanecek **(Technology Centre AS CR, Czech Republic)**
Karen D. Sermon **(ESHRE)**
Eleni Zika, Dolores Ibarreta **(JRC - IPTS)**

Services and Centres

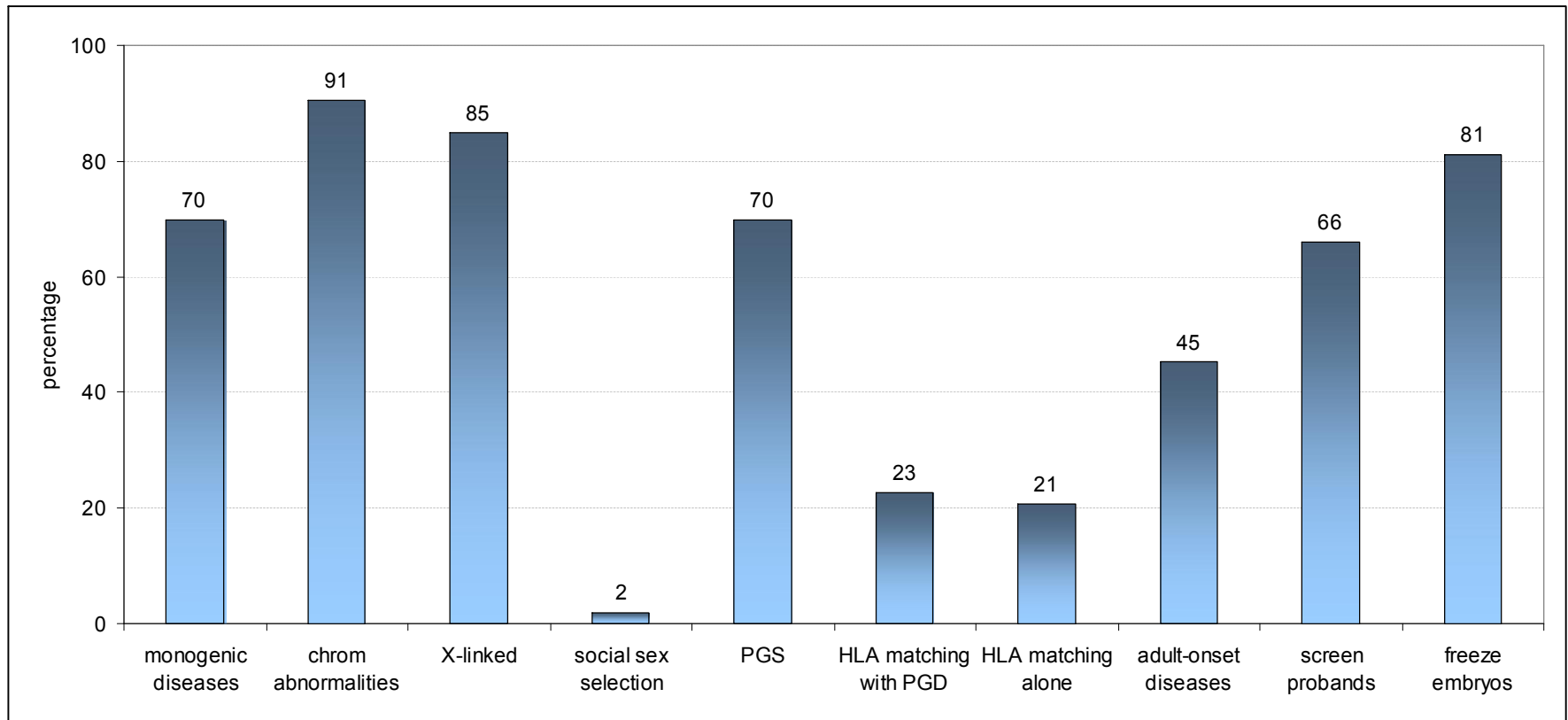
Overall distribution of services provided



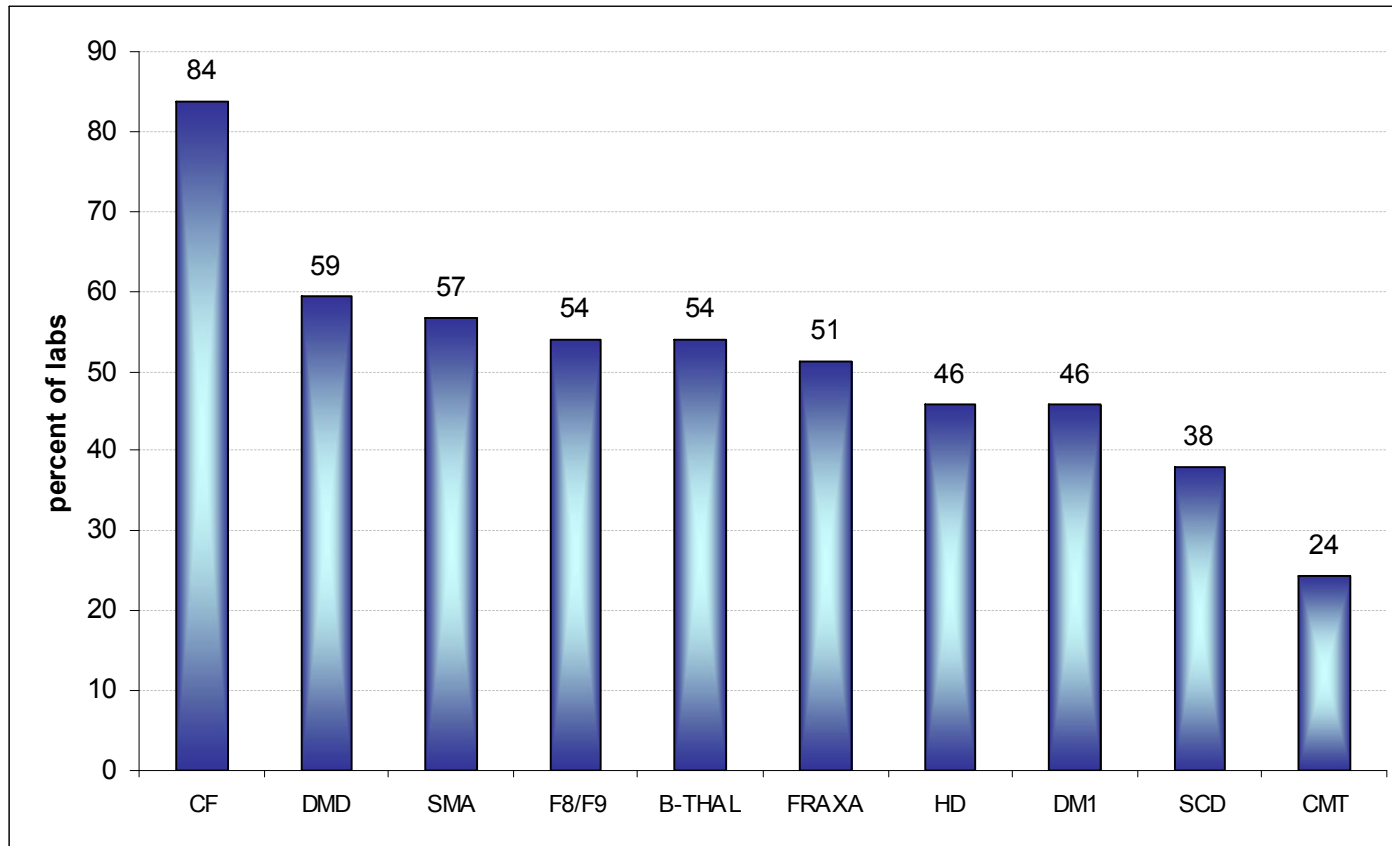
Affiliation of labs and clinics



Services available in the centres offering PGD



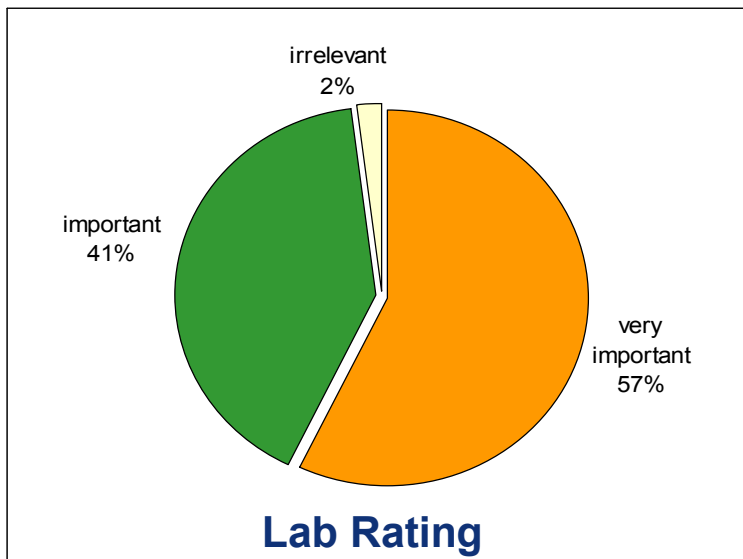
Availability of testing for 10 common monogenic diseases



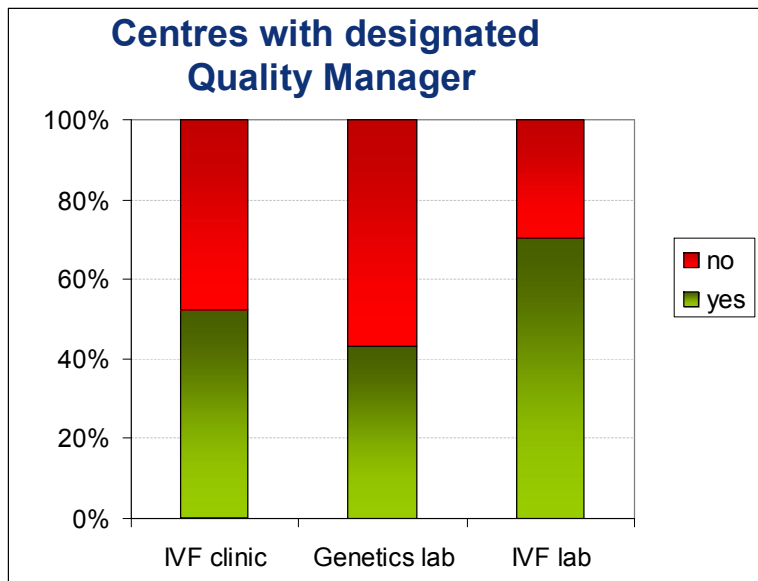
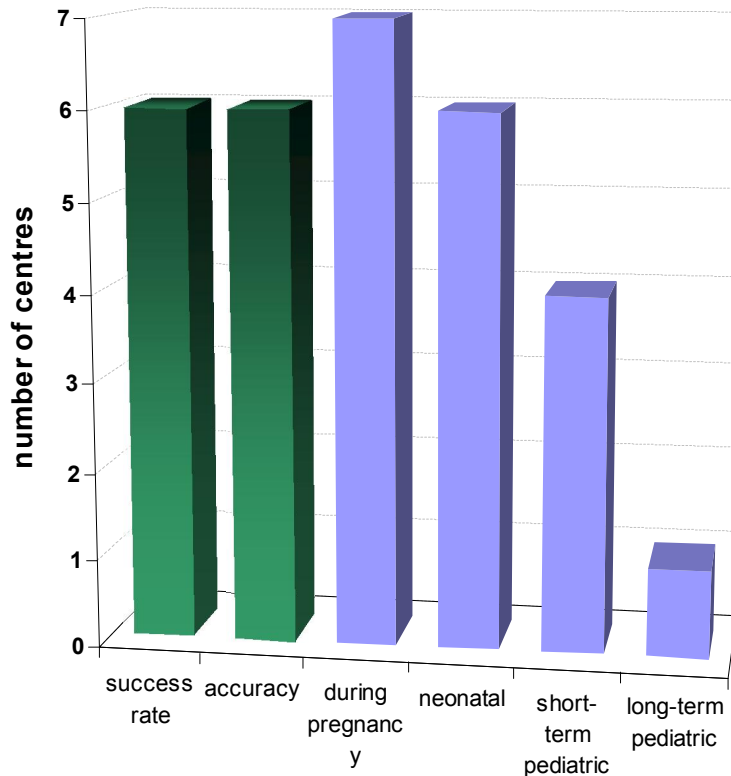
Detailed results by country:

	CF	DM	DSMA	F8/F9	β THAL	FRAXA	HD	DM1	SCD	CMT
Belgium [n = 6]	3	3	3	1	2	2	2	2	3	2
Cyprus [1]	1	1		1	1		1			
Czech Republic [6]	2	1	1	1		1				
Denmark [1]	1	1				1	1	1		
Finland [2]						1		1		
France [3]	3	3	3	2	2	2	2	3	2	1
Germany [3]	1	1		1		1		1		
Greece [5]	4	1	1	2	5	1	1	1	3	
Portugal [1]				1			1			
Spain [8]	7	6	5	5	3	5	5	3	2	3
Sweden [2]	1	1	2	1	1	1		1		
Switzerland [1]	1				1					
Netherlands [3]	1		1			1	1	1		
Turkey [2]	2	2	2	2	2	2	1	1	2	2
UK [5]	4	2	2	2	1		2	1	1	

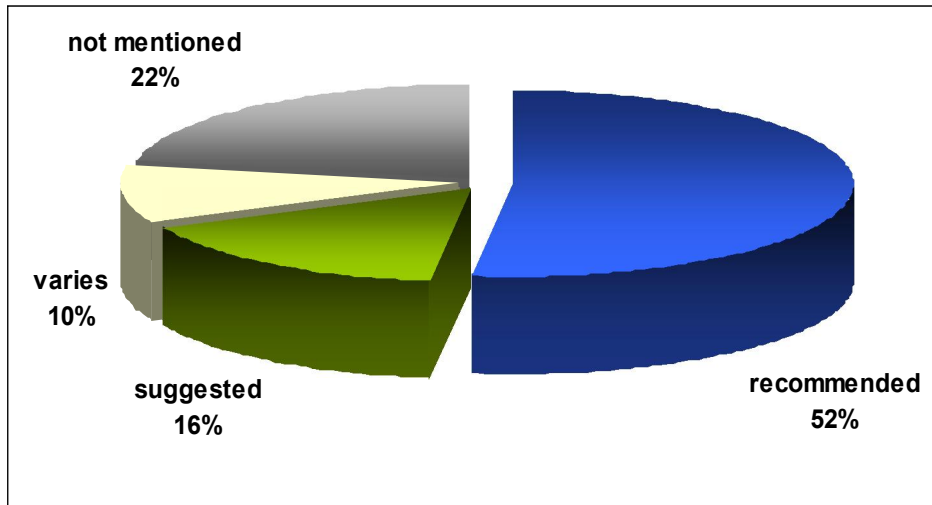
Quality Assurance



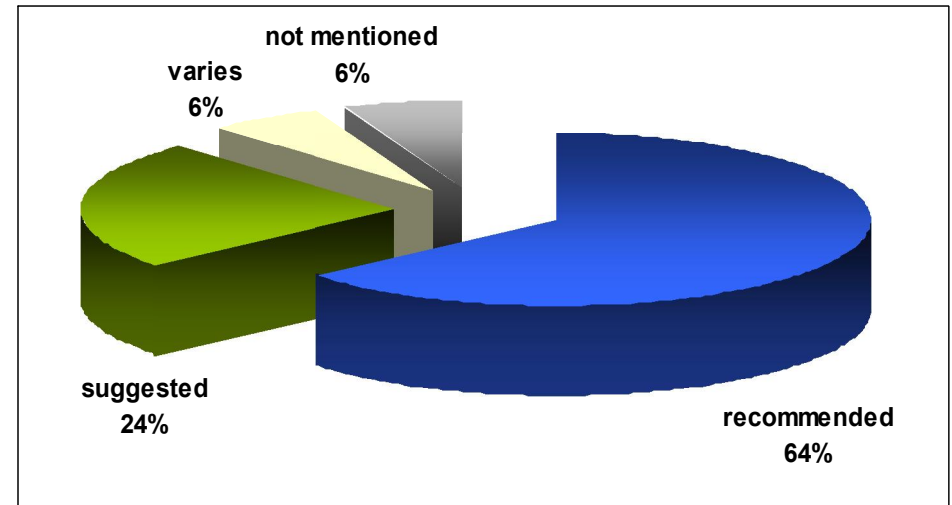
Follow-up of outcomes



Recommendation for confirmation of diagnosis

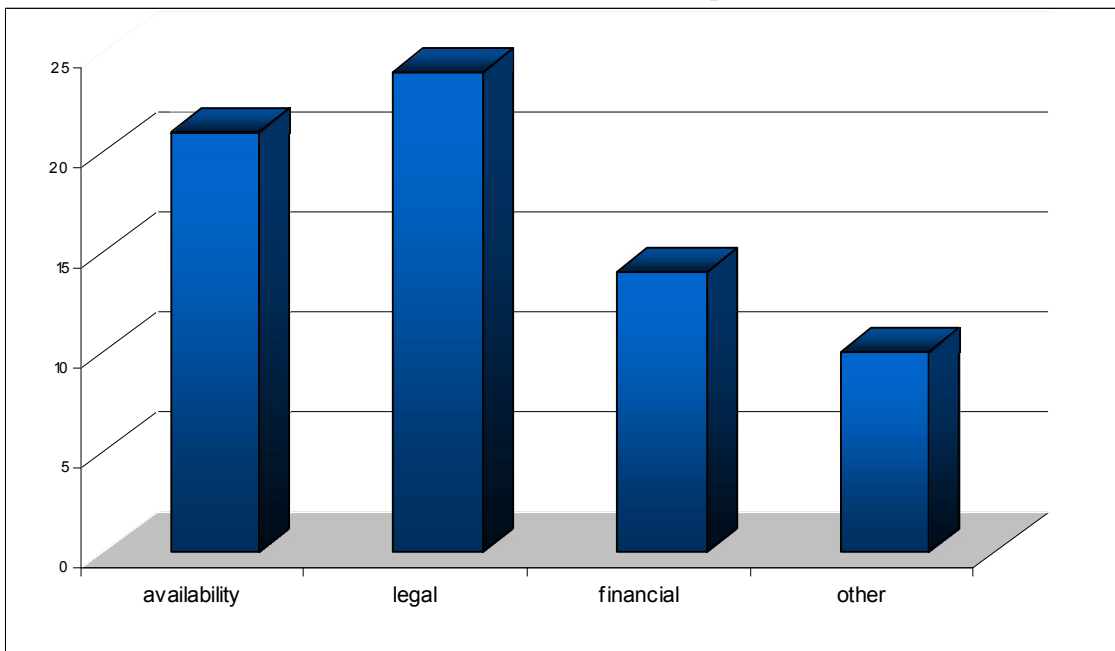


Reports



Informed consent

Trans - border flow of patients



Patient's reasons



Receiving centres and patients

	Number of centers that treat patients from abroad	Number of patients
Spain	8	332
Cyprus	1	150
Belgium	6	127
Czech Republic	6	110
Turkey	3	35
Germany	3	22
Slovakia	1	20
Greece	6	18
France	3	10
The Netherlands	3	2
United Kingdom	5	-
other	8	6
Total	53	832

Country	Abortion	PND	PGD
Belgium	Allowed, subject to conditions. After 14 weeks, only allowed if mother's life in danger, or if clear that child will suffer from an incurable disease.	Allowed	No specific regulation but existing regulations for ART influence PGD.
Czech Republic	Allowed, subject to conditions. After 12 weeks, only allowed if mother's life is endangered or where there is risk of serious damage to or 'life incapability' of the embryo. In cases of genetic indications, allowed up to 24 weeks.	Allowed and not regulated	Allowed for specified indications and only to exclude the risk of serious genetic diseases.
France	Allowed up to 12 weeks. Allowed after 12 weeks where high probability of birth of affected child with a severe disease recognised as incurable.	Allowed subject to regulation.	Allowed in certain circumstances and subject to regulation and licensing
Germany	Allowed. After 14 weeks, no time limit if for medical reasons	Allowed and not regulated	Not allowed except for polar body analysis.
Ireland	Not allowed	Allowed and not regulated	Not allowed
Netherlands	Allowed up to 24 weeks.	Allowed and self regulated.	Allowed and regulated.
Slovakia	Allowed up to 12 weeks. After 12 weeks, only allowed where risk to mother's life or risk of serious damage or 'life incapability' of the embryo.	Allowed and not currently regulated but guidance through ordinance of Ministry of Health.	Allowed and not currently regulated but draft law prepared.
Spain	Allowed where: 1. severe risk for the life or physical or psychological health of pregnant woman; 2. 'woman distress'; and 3. presumption that foetus may be born with severe physical or mental defects.	Allowed and not regulated.	Allowed subject to legislation which allows PGD for severe genetic disorders or as therapeutic tool for existing affected children.
Switzerland	Allowed up to 12 weeks on written request and in a case of "a state of distress" After 12 weeks, requires medical opinion required, indicating abortion necessary to avoid danger of serious threat to physical integrity or of a state of severe distress of pregnant woman.	Allowed subject to some regulation. Determination of characteristics that do not directly affect health of the embryo or foetus, and social sex selection prohibited.	Not allowed except for polar body analysis.
United Kingdom	Allowed in specific circumstances defined by legislation.	Allowed and not regulated.	Allowed subject to regulation, licensing and inspection.

1. *PGD is an expanding activity in Europe with increasing social implications (e.g. trans-border flow of couples)*
2. *There is a diversity of tests available but **trend towards custom-made tests**, most commonly for extremely rare disorders*
3. *Type of tests performed shows a different perception of PGD – used in some indications which may be less acceptable in prenatal diagnosis*
4. *Questionnaire results suggest that **formal EQA schemes are yet to be implemented** in the majority of the centres*



*Simultaneous first and second polar bodies removal,
Photo: V. Ivakhenko*

5. *Need for development of PGD-specific **quality assurance schemes** (or adaptation of existing ones)*
6. ***Genetic counselling and informed consent guidelines**, adapted for PGD need to be developed*
7. *The **EU Human Tissue and Cells Directive** will positively impact quality standards but requires technical specificities for PGD*
8. *There is a **need to support monitoring** of PGD treatment, with increased public funding and international co-operation.*



9. *Different regulatory frameworks across countries creates cross-border flow of patients.*

10. ***Trans-border flow:** approximately one third of all the cycles identified by the survey are referred internationally. Main reasons: legal (i.e. PGD not allowed in the country of residence) others (**quality** of the treatment, test **availability**, financial **resources** and **man-power**)*

***Potential disadvantages:** Problems with **referral** and difficulties with the **monitoring and follow-up** (impaired when couples go back to their countries).*



Thank you very much for your attention

The *Preimplantation Genetic Diagnosis* study can be downloaded free from:

<http://ipts.jrc.ec.europa.eu>

The EU Tissue and Cells Directive 2004/23/EC and its implementing technical directives can be downloaded from:

http://ec.europa.eu/health/ph_threats/human_substance/legal_tissues_cells_en.htm