Pwyllgor Gwasanaethau Iechyd Arbenigol Cymru (PGIAC)
Welsh Health Specialised Services Committee (WHSSC)

Specialised Services Policy:

CP37
Pre-implantation Genetic Diagnosis (PGD)

Document Author:	Specialised Planner, Women & Children
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Document History

Revision History			
Version No.	Revision date	Summary of Changes	Updated to version no.:
3.1	January 2013	• •Paragraph added section 3.2 'If it is decided after an unsuccessful cycle, that the treatment is unlikely to benefit the couple, further treatment should not be offered'. • Section 3.3, change to Referral Pathway preferred provider bring Guy's & St Thomas in the first instance. • 3.4 Exclusions 'unless chromosomal abnormality detected'. • 3.3 Referral Pathway - text added to say that PGD assessment will be funded in the first instance only for patients who meet the access criteria. • 3.7 Governance arrangements added. • Audit Arrangements added. • Referral Pathway Flowchart updated. • Policy transferred to new template.	
3.1	05/03/2013	Ratified through Chair's Action on behalf of Management Group	4.0
4.0	March 2014	Language amended to be clearer and more consistent Definition of a cycle expanded in line with the Specialist Fertility Policy Criteria made more explicit instead of referring to appendix and consistent with the Specialist Fertility Policy Referral pathway made more explicit	4.1
4.1	June 2014	Slight amendments to language following consultation with stakeholders	4.2
4.2	05/08/2014	Approved by Executive Board. Ratified through Chair's Action on behalf of Management Group	5.0
Date of next revision			

Consultation

Name	Date of Issue	Version
All Wales Medical Genetics Service	April 2014	
All Wales Specialist Fertility Advisory Group	April 2014	
PGD Provider	April 2014	
WHSSC Women and Children's Programme Team	May 2014	

Approvals		
Name	Date of Issue	Version No.
HCW Commissioning Business Meeting	20/08/08	0.1
All Wales Specialist Fertility Advisory Group	08/09/08	0.2
All Wales Specialist Fertility Advisory Group	17/12/08	0.3
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WHSSC Executive Team		2.0
WHSSC Joint Committee		2.0
WHSSC Women and Children's Programme Team	28/07/11	3.0
WHSSC Management Group (Chair's action)	05/03/13	4.0
WHSSC Women and Children's Programme Team	May 2014	4.2
WHSSC Management Group (Chair's action)	05/08/14	5.0

Distribution – *this document has been distributed to*			
Name	By	Date of Issue	Version No.
Providers			
LHB Medical Directors			

Policy Statement

Background	Pre-implantation genetic testing is a technique used in reproductive medicine to identify genetics defects in embryos created through in vitro fertilisation (IVF). PGD can be offered when one or both genetic parents have, or are carriers of, a known genetic abnormality. Testing is performed on their embryos to determine whether the embryo is at risk of genetic disease. Unaffected embryos are selected for transfer back to the uterus in the hope that a normal birth will ensue.
Summary of clinical criteria	Patients who meet the access criteria are entitled to up to three full cycles of PGD at the preferred providers outlined in the Policy. The circumstances of those requesting PGD may be different to those requesting IVF for infertility reasons, and as a result the application of some the IVF access criteria in place for Welsh patients is inappropriate for PGD. In order to access PGD the following criteria should be met: • The couple should be at risk of having a child with a serious genetic condition and this risk must be greater than 10%; • Each case must have had the appropriate guidance and advice from the All Wales Medical Genetics Service and each couple should have received genetic counselling from a clinical geneticist or a registered genetic counsellor; • Both partners should be non smokers at time of treatment • Both partners should have a body mass index of between at least 19 and up to and including 30; • The female partner should be under 43 years of age at the time of treatment; • Couples who have a chromosome rearrangement will often carry a risk of less than 10% of having an affected live-born baby. However, for these couples

	who have a documented inheritable chromosome rearrangement PGD may be considered in the following circumstances: - When a woman has experienced / or may experience a late termination of pregnancy - There has been a history of still births in an affected pregnancy - There is a history of recurrent miscarriages associated with a documented chromosome rearrangement; this will exclude sporadic standard aneuploidies which can occur with increasing maternal age. • A woman shall not receive treatment unless account has been taken of the welfare of any child who may be born as a result; • Patients should conform with the Human Fertilisation and Embryology Authority (HFEA) Code of Practice; • Neither partner has undergone sterilisation (this does include conditions where sterilisation occurs as a result of another medical problem); • There should be no living unaffected children from the current relationship; • The genetic condition is licensed by the HFEA, and is listed in its website (Annex (iv)); • The test must be included in the list of UKGTN approved tests, or suitable for inclusion; In addition to technological limitations, standards exist that specify what PGD can and cannot be used for. At the UK level, the HFEA closely monitors the application of the procedure. With regard to PGD, for example, it states that its use is prohibited in sex selection for non-medical reasons.

Responsibilities	Referrers should: • Inform the patient that this treatment is not routinely funded outside the criteria in this policy; and • Refer via the agreed pathway. Clinicians considering treatment should: • Discuss all the alternative treatment with the patient; • Advise the patient of any side effect and risks of the potential treatment; • Inform the patient that treatment is not routinely funded outside of the criteria in the policy; and • Confirm that there is contractual agreement with WHSSC for the treatment. If the patients does not meet the access criteria and there are exceptional circumstances, the clinician should submit an IPFR request.

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1. Aim

1.1 Introduction

The document has been developed as the policy for the planning of Pre-implantation Genetic Diagnosis for Welsh patients. The policy applies to residents of all seven Health Boards in Wales.

The purpose of this document is to:

- Set out the circumstances under which patients will be able to access Pre-implantation Genetic Diagnosis services;
- clarify the referral process and;
- define the criteria that patients must meet in order to access treatment.

1.2 Relationship with other Policies and Service Specifications

This document should be read in conjunction with the following documents:

- Specialised Services Policy for Specialist Fertility Services; and
- All Wales Policy: Making Decisions on Individual Patient Funding Requests (IPFR)

2. Scope

2.1 Definition

Pre-Implantation Genetic Diagnosis is a technique that involves testing cell(s) from embryos created outside the body by *in vitro* fertilisation (IVF) for a genetic disorder. The tests are carried out for a specific disorder that is known to be present in the family and from which the embryos are known to be at significant risk. Unaffected embryos are selected for transfer back to the uterus in the hope that a normal birth will ensue.

Patients who meet the NHS access criteria are entitled to receive up to three complete cycles of PGD. A completed PGD cycle should comprise of ovarian stimulation; egg recovery; egg recovery; fertilisation; embryo biopsy; genetic testing and the transfer of any resultant fresh and frozen embryo(s). This will include the storage of any frozen embryos for one year following egg collection.

Patients will need to be advised at the start of the treatment that this is the level of service that is available on the NHS and that the NHS will not fund storage following this period. Patients who have frozen embryos remaining after their first fresh cycle should utilise the previously frozen embryos, rather than undergo ovarian stimulation, egg retrieval, and fertilization again. Frozen embryos must be transferred within ten years of the initial treatment cycle (HFEA guidance).

The circumstances of those requesting PGD may be different to those requesting IVF for infertility reasons, and as a result the application of some of the IVF access criteria in place for Welsh patients is inappropriate for PGD.

2.2 Codes

ICD-10 code	Description
Z31.2	In vitro fertilization
Z31.3	Other assisted fertilization methods

OPCS-4.6 code	Description
Q13	Introduction of gamete into uterine cavity
Q13.1*	Transfer of embryo to uterus NEC
Q21	Other introduction of gamete into uterine cavity
Q21.1*	Transmyometrial transfer of embryo to uterus
* Note: Q13.1 and Q21.1 will require the following code from Y96.- (below) to identify the PGD (but the code from Y96 will never appear *without* one of the above two codes)	
Y96	In vitro fertilisation
Y96.5	In vitro fertilisation with pre-implantation for genetic diagnosis

3. Access Criteria

3.1 Clinical Indications

PGD offers couples (who are not necessarily infertile) the opportunity of having a healthy child of their own, whilst avoiding having to undergo a termination of an affected fetus(es) detected through prenatal diagnosis (for example, amniocentesis). For many people, a termination is either unacceptable or less preferable.

Indications for PGD are where a couple is at risk of transmitting serious genetic disorders to their offspring and this risk is greater than 10%. The couple may not necessarily have infertility issues.

It is acknowledged that choosing to proceed with PGD is not an easy option and couples who could be regarded as being in need may not necessarily proceed with treatment following assessment.

3.2 Criteria for Treatment

Patients who meet the NHS access criteria are entitled to receive up to three complete cycles of PGD. A completed PGD cycle should comprise of ovarian stimulation; egg recovery; egg recovery; fertilisation; embryo biopsy; genetic testing and the transfer of any resultant fresh and frozen embryo(s). This will include the storage of any frozen embryos for one year following egg collection. Patients who have frozen embryos remaining after their fresh cycle should utilise these frozen embryos first, rather than undergo ovarian stimulation, egg retrieval, and fertilization again.

Couples who have previously self funded PGD will be entitled to NHS treatment to reach up to the three completed cycles. Where patients have frozen embryos remaining after a self funded cycle that has not lead to pregnancy, these should be utilised first before proceeding with ovarian stimulation, egg retrieval, and fertilization again.

The circumstances of those requesting PGD may be different to those requesting IVF for infertility reasons, and as a result the application of some of the IVF access criteria in place for Welsh patients is inappropriate for PGD.

In order to access PGD the following criteria must be met:

- The couple should be at risk of having a child with a serious genetic condition and this risk must be greater than 10%;
- Each case must have had the appropriate guidance and advice from the All Wales Medical Genetics Service and each couple should have received genetic counselling from a clinical geneticist or a registered genetic counsellor*;*
- Both partners should be non smokers at time of treatment
- Both partners should have a body mass index of between at least 19 and up to and including 30;
- The female partner should be under 43 years of age at the time of treatment. Where the female partner is aged less than 40 and meet the criteria they will be entitled to up to three cycles of PGD. Where the female partner is aged between 40 and 42 and meet the criteria they will be entitled to one cycle of PGD as long as they meet the following criteria:
    - They have never previously had PGD treatment
    - There is no evidence of low ovarian reserve
    - There has been a discussion of the additional implications of PGD and pregnancy at this age
- Couples who have a chromosome rearrangement will often carry a risk of less than 10% of having an affected live-born baby. However, for these couples who have a documented inheritable chromosome rearrangement PGD may be considered in the following circumstances:
    - When a woman has experienced / or may experience a late termination of pregnancy
    - There has been a history of still births in an affected pregnancy
    - There is a history of recurrent miscarriages associated with a documented chromosome rearrangement; this will exclude sporadic standard aneuploidies which can occur with increasing maternal age.
- A woman shall not receive treatment unless account has been taken of the welfare of any child who may be born as a result;
- Patients should conform with the Human Fertilisation and Embryology Authority (HFEA) Code of Practice;
- Neither partner has undergone sterilisation (this does include conditions where sterilisation occurs as a result of another medical problem);
- There should be no living unaffected children from the current relationship;

- The genetic condition is licensed by the HFEA, and is listed in its website (Annex (iv));
- The test must be included in the list of UKGTN approved tests, or suitable for inclusion;

If it is decided after an unsuccessful cycle, that the treatment is unlikely to benefit the couple, further treatment should not be offered.

3.3 Referral Pathway

Each case should be assessed and considered individually. Patients must be referred for discussion and genetic counselling advice to the All Wales Medical Genetics Service (AWMGS) by their secondary care consultant or GP once all appropriate testing and investigations have been undertaken.

WHSSC recognises that the need for PGD may often be identified by the couple following investigation into their family history or via other routes, it is therefore reasonable for a GP to make a referral to the AWMGS for further advice.

The AWMGS will act as a source of advice for patients. Once the need for PGD has been established and their suitability and eligibility have been assessed and agreed by AWMGS; the WHSSC Assessment Form should be completed by the AWMGS. The Assessment Form is attached as Annex (i). AWMGS will ensure that a copy of the assessment form is provided to the appropriate patient's referrer (e.g. GP) with guidance on the next steps for the application.

An application will then be made to WHSSC by the referrer using the Checklist Form in Annex (iii) with the WHSSC Assessment Form. As long as the couple meet the criteria and the WHSSC Assessment Form supports PGD, WHSSC will provide prior approval for funding for PGD assessment at the designated centre in the first instance.

WHSSC will consider approval of funding for PGD assessment at a designated centre in the first instance for couples who meet the access criteria. An initial outpatient appointment will be made for the clinic to assess the couple and to discuss whether PGD might be possible.

Following this assessment if applicable and the couple wish to proceed to full PGD treatment then the designated centre will

complete a further funding application request and forward to WHSSC for consideration and approval.

It is the responsibility of the AWMGS and the PGD provider to ensure that the couple meets all areas of the access criteria

The designated provider for PGD is:

- Assisted Conception Unit, Guy's & St Thomas Hospital

If Guy's and St Thomas Hospital is unable to undertake a PGD test for a particular couple because they are not licensed by the HFEA for the test then Welsh Health Specialised Services will consider funding treatment under exceptional circumstances at the following provider:-

- Assisted Conception Unit, University College London Hospital

If the patient wishes to be referred to a provider outside of the agreed pathway, an IPFR can be submitted if there are exceptional circumstances.

3.4 Exclusions

In addition to technological limitations, standards exist that specify what PGD can and cannot be used for. At the UK national level the HFEA closely monitors the application of the procedure. Clinics have to adhere to the HFEA's Code of Practice, which gives guidance on the proper conduct of licensed activities of techniques for assisted reproduction.

The following uses of the PGD technology are excluded from this policy.

- Non medical gender selection e.g. for the purpose of family balancing. This is illegal in the United Kingdom (UK)
- Human Leucocyte Antigen (HLA) typing to produce a donor sibling for a child requiring an allogeneic stem cell transplant.
- Using PGD to address infertility or to prevent miscarriages of unknown aetiology
- Pre-implantation Genetic Screening (PGS). Here, genetic testing is used to screen embryos for various abnormalities in chromosomes typically the number of chromosomes (chromosomal aneuploidies)

3.5 Exceptions

If the patient does not meet the criteria for treatment, but the referring clinician believes that there are exceptional clinical grounds for treatment an Individual Patient Funding Request (IPFR) can be made to WHSS under the All Wales Policy for Making Decisions on Individual Patient Funding Requests (IPFR).

If the patient wishes to be referred to a provider out of the agreed pathway, an IPFR should be submitted.

Guidance on the IPFR process is available at www.whssc.wales.nhs.uk

3.6 Responsibilities

Referrers should:

- Fairly inform patients of their eligibility of treatment under this Policy;
- Inform the patient that this treatment is not routinely funded outside the criteria in this policy; and
- Refer via the agreed pathway.

Clinicians considering treatment should:

- Discuss all the alternative treatment with the patient;
- Advise the patient of any side effect and risks of the potential treatment;
- Inform the patient that treatment is not routinely funded outside of the criteria in the policy; and
- Confirm that there is contractual agreement with WHSSC for the treatment.

If a patient does not meet the access criteria and there are exceptional clinical circumstances, the clinician should submit an IPFR request.

3.7 Governance Arrangements

WHSSC expects robust mechanisms to be put in place to support the clinical governance of providers and to ensure these comply with the HFEA Code of Practice including:

1. The centre must have a valid HFEA licence which includes the

provision of PGD, and abide by the HFEA regulations for PGD testing. WHSSC will only purchase PGD services from PGD providers who have been licensed by the HFEA. WHSSC will monitor reports provided by the HFEA and will discuss the findings with the provider where appropriate.

2. The laboratory where the test is being carried out must have Clinical Pathology Accreditation (CPA).

3. There must be an existing licence to carry out a specific test from the HFEA or PGD clinic must apply for and receive a licence prior to treatment if that condition is not currently licensed.

In addition to the approval of the HFEA, clinics must make their own judgement about whether PGD is appropriate treatment for a particular couple, using guidance contained in the HFEA's Code of Practice.

3.8 Audit Arrangements

As part of this policy PGD service providers will provide two sets of auditable data to WHSSC for all the NHS PGD cycles they have provided:

Data set 1: Monthly minimum data set on the PGD cycles completed for WHSSC patients for that month.

Data set 2 : An annual report and dataset on all patients who were referred to the PGD provide. This annual report should provide information on number of patients that attended for assessment and those that proceeded for treatment as well as the desired primary outcome of PGD which is the birth of an unaffected baby. In addition to the live birth rates, reports should also cover secondary indicators such as:-

- Multiple Births
- Prematurity and low birth weight
- Method of Delivery
- Misdiagnosis
- Other complications
- Acceptance of the procedures

This data and reports would not only enable monitoring of this PGD policy but would also enable outcome information to be collated to inform future PGD policy needs.

4. Putting Things Right: Raising a Concern

Whilst every effort has been made to ensure that decisions made under this policy are robust and appropriate for the patient group, it is acknowledged that there may be occasions when the patient or their representative are not happy with decisions made or the treatment provided. The patient or their representative should be guided by the clinician, or the member of NHS staff with whom the concern is raised, to the appropriate arrangements for management of their concern:

- When a patient or their representative is unhappy with the decision that the patient does not meet the criteria for treatment further information can be provided demonstrating exceptionality. The request will then be considered by the All Wales IPFR Panel.
- If the patient or their representative is not happy with the decision of the All Wales IPFR Panel the patient and/or their representative has a right to ask for this decision to be reviewed. The grounds for the review, which are detailed in the All Wales Policy: Making Decisions on Individual Patient Funding Requests (IPFR), must be clearly stated. The review should be undertaken, by the patient's Local Health Board;
- When a patient or their representative in unhappy with the care provided during the treatment or the clinical decision to withdraw treatment provided under this policy, the patient and/or their representative should be guided to the LHB for NHS Putting Things Right. For services provided outside NHS Wales the patient or their representative should be guided to the NHS trust Concerns Procedure, with a copy of the concern being sent to WHSSC.

5. Equality Impact and Assessment

The Equality Impact Assessment (EQIA) process has been developed to help promote fair and equal treatment in the delivery of health services. It aims to enable Welsh Health Specialised Services Committee to identify and eliminate detrimental treatment caused by the adverse impact of health service policies upon groups and individuals for reasons of race, gender re-assignment, disability, sex, sexual orientation, age, religion and belief, marriage and civil partnership, pregnancy and maternity and language (welsh).

This policy has been subjected to an Equality Impact Assessment. The Assessment demonstrates the policy is robust and there is no potential for discrimination or adverse impact. All opportunities to promote equality have been taken.

Annex (i) WHSSC Assessment Form

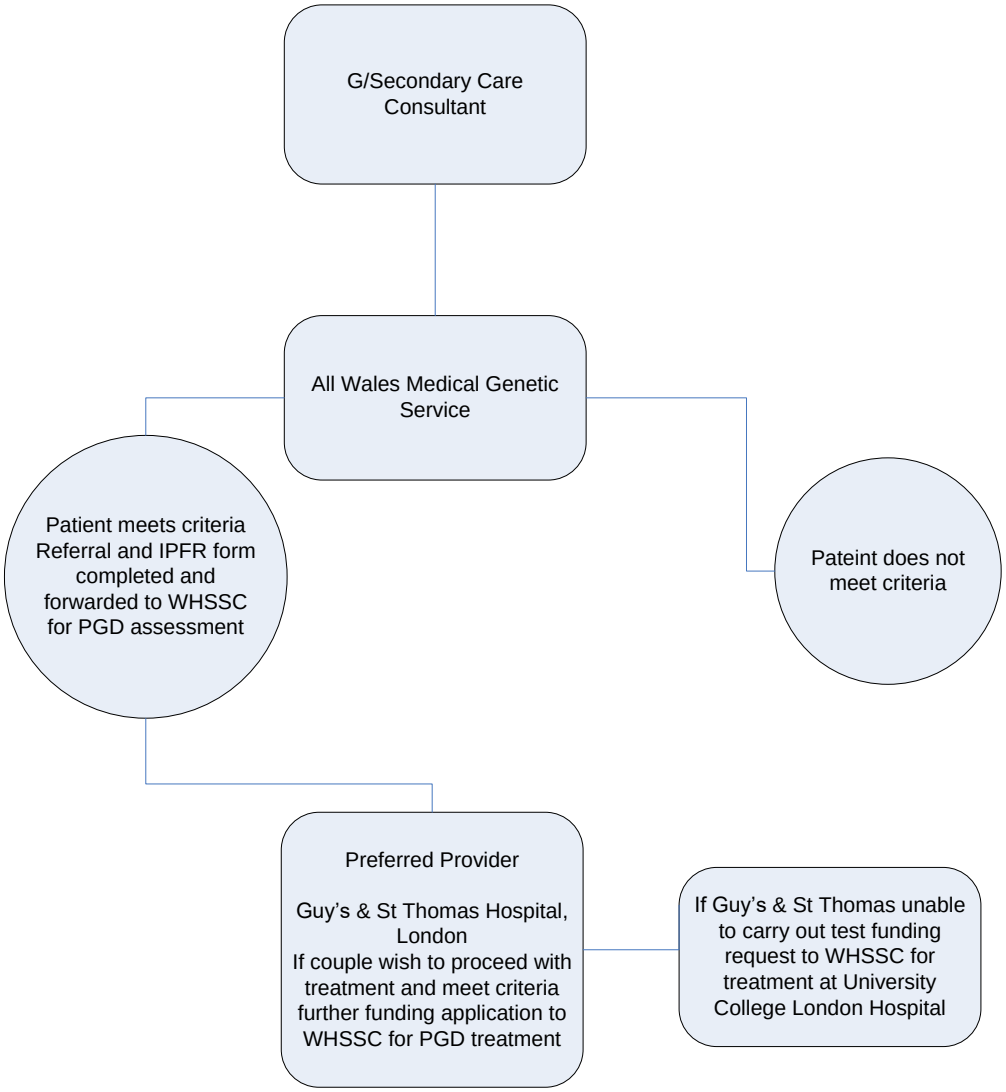
WHSSC Assessment Form to be completed by AWMGS
PREIMPLANTATION GENETIC DIAGNOSIS

Completed By:		
Please tick box or write in details where indicated.		
1. Patient's initials:		**NHS Number:**
2. Date of referral:		
3. Patient's Date of Birth:		
4. Patient's Post Code:		
5. Referrer's Name and Address:		
6. Reason for referral (please tick):		
- Previous child born with disorder:		
- Patient/partner has disorder:		
- Known family history of disorder:		
- Other		Please specify:
7. Disorder(s) being tested/treated for:		

	Please Tick
8. The above disorder is a condition licensed by the HFEA:	
9. Account has been taken of the welfare of any child who may be born as a result:	
10. The couple meet the criteria as set out in 3.2 of the WHSSC PGD Policy:	

11. Couple's status:	
- Not suitable for PGD/not eligible:	
- Eligible for PGD but decided not to proceed further:	
- Eligible for PGD and happy to proceed:	
- Eligible but received or currently receiving treatment (please specify):	
I can confirm that the eligibility criteria guidance in 3.2 of the CP37 PGD policy has been fully considered.	
Signed:	
Date:	

Annex (ii) Referral Pathway



Annex (iii) Checklist

Pre-Implantation Genetic Diagnosis

The following checklist should be completed and retained as evidence of policy compliance by the receiving centre. It is expected that this evidence will be provided at the point of invoicing by the receiving centre.

i) Where the patient meets the criteria **AND** the procedure is included in the contract **AND** the referral is received by an agreed centre, the form should be completed and retained by the receiving centre for audit purposes.
ii) The patient meets the criteria **AND** is received at an agreed centre, but the procedure is not included in the contract. The checklist must be completed and submitted to WHSSC for prior approval to treatment.
iii) The patient meets the criteria but wishes to be referred to a non contracted provider. An Individual Patient Funding Request (IPFR) Form must be completed and submitted to WHSSC for consideration.
iv) The patient does not meet criteria, but there is evidence of exceptionality. An Individual Patient Funding Request (IPFR) Form must be completed and submitted to WHSSC for consideration for treatment.

To be completed by the referring gatekeeper or treating clinician and sent to WHSSC with the Assessment Form in Annex (i)

PRIOR APPROVAL FOR PGD

Patient NHS No:				
Patient is Welsh Resident	Post Code			
Patient is English Resident	GP Code:			
Patient meets following access criteria for treatment (please tick):			**Yes**	**No**
The couple should be at risk of having a child with a serious genetic condition and this risk must be greater than 10%				
Each case must have had the appropriate guidance and advice from the All Wales Medical Genetics Service and each couple should have received genetic counselling from a clinical geneticist or a registered genetic counsellor				
Both partners should be non smokers at time of treatment				
The female partner should be under 43 years of age at the time of treatment				
Couples who have a chromosome rearrangement will often carry a risk of less than 10% of having an affected live-born baby. However, for these couples who have a documented inheritable chromosome rearrangement PGD may be considered in the following circumstances: - When a woman has experienced / or may experience a late termination of pregnancy - There has been a history of still births in an affected pregnancy - There is a history of recurrent miscarriages associated with a documented chromosome rearrangement; this will exclude sporadic standard aneuploidies which can occur with increasing maternal age.				
A woman shall not receive treatment unless account has been taken of the welfare of any child who may be born as a result;				
Patients should conform with the Human Fertilisation and Embryology Authority (HFEA) Code of Practice;				
Neither partner has undergone sterilisation (this does include conditions where sterilisation occurs as a result of another medical problem);				
There should be no living unaffected children from the current relationship;				
The genetic condition is licensed by the HFEA, and is listed in its website (Annex (iv));				
The test must be included in the list of UKGTN approved tests, or suitable for inclusion;				
Patient wishes to be referred to non-contracted provider				
An Individual Patient Funding Request (IPFR) must be completed and submitted to WHSSC for approval prior to treatment. The form must clearly demonstrate why funding should be provided as an exception. The form can be found at http://www.wales.nhs.uk/sites3/docopen.cfm?orgid=898&id=181455				
Patient does not meet access criteria but is exceptional				
An Individual Patient Funding Request (IPFR) must be completed and submitted to WHSSC for approval prior to treatment. The form must clearly demonstrate why funding should be provided as an exception. The form can be found at http://www.wales.nhs.uk/sites3/docopen.cfm?orgid=898&id=181455				

Name: _____ **Designation:** _____

Signature: _____ **Date:** _____

	Name (printed):	Signature:	Date:	Yes	No
Authorised by TRM Gatekeeper					
Authorised by Patient Care Team					
Authorised by Agreed other (Please state)					
Patient Care Team/IPFR TRM Reference number:					

Annex (iv) Conditions Licensed

PGD is an area of medicine that is rapidly developing, and new tests often become available. If a condition does not appear on this list, we recommend reviewing the HFEA website or getting in touch with a licensed PGD clinic.

Conditions licensed
(PIGN gene) Multiple Congenital Anomalies Hypotonia – Seizures Syndrome 1
5 Alpha Reductase Deficiency (5ARD) insofar as that condition affects males, with simultaneous sex determination
Acute Intermittent Porphyria
Acute Recurrent Autosomal Recessive Rhabdomyolysis (ARARRM)
Adrenoleukodystrophy (Adrenomyeloneuropathy)
Agammaglobulinaemia
Aicardi Goutieres Syndrome 1 (AGS1)
Alagille Syndrome
Alpers Syndrome
Alpha-1-antitrypsin deficiency
Alpha-mannosidosis
Alpha thalassaemia/mental retardation syndrome*
Alpha Thalassemia
Alports Syndrome
Alports Syndrome (Autosomal Dominant)
Alzheimers Disease - early onset
Amyotrophic Lateral Sclerosis 1 (ALS1)
Anderson Fabry Disease
Androgen Insensitivity Syndrome
Angelman Syndrome (UBE3A gene only)
Aplastic anaemia - severe*
Argininosuccinic Aciduria
Arrhythmogenic Right Ventricular Cardiomyopathy/ Dysplasia (ARVC/D), Autosomal Dominant
Ataxia Telangiectasia
Autosomal Dominant Acute Necrotizing Encephalopathy
Autosomal Dominant Polycystic Kidney Disease (ADPKD)
Autosomal Dominant Retinitis Pigmentosa
Autosomal Dominant Retinitis Pigmentosa
Autosomal Recessive Dopa Responsive Dystonia
Autosomal Recessive Severe Combined Immunodeficiency with Bilateral Sensorineural Deafness
Bardet-Biedl syndrome (BBS)
Barth Syndrome
Battens Disease (infantile)

Beta Hydroxyisobutyryl CoA Hydrolase Deficiency (Methacrylic Aciduria)
Beta Thalassaemia*
Bethlem Myopathy
Bilateral Frontoparietal Polymicrogyria
Birt-Hogg-Dubé Syndrome
Branchio-Oto-Renal Syndrome (BOR)
BRCA 1 (increased susceptibility to breast cancer)
Breast Ovarian Cancer Familial Susceptibility (BRCA2)
Bruton Agammaglobulinemia Tyrosine Kinase (BTK)
Calpainopathy
Canavan Disease
Cardiac Valvular Dysplasia
Carney Complex
Carnitine Acylcarnitine Translocase Deficiency (CACT)
Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT1)
Catecholaminergic Polymorphic Ventricular Tachychardia 2 (CPVT2)
Central Core Disease of Muscle
Cerebral Autosomal Dominant Arteriopathy with Sub cortical infarcts and Leukoencephalopathy (CADASIL)
Cerebral Cavernous Malformations (CCM)
Charcot Marie Tooth Disease
Charcot Marie Tooth Disease Type 2
Charcot Marie Tooth Disease, demyelinating, type 1A (CMT1A)
Chondrodysplasia Punctata
Choroideraemia
Chromosomal rearrangements (various)
Chronic Granulomatous Disease
Citrullinaemia type 1
Classical Ehlers Danlos Syndrome
Coffin-Lowry Syndrome
Cohen Syndrome
Congenital Adrenal Hyperplasia (21 hydroxylase deficiency)
Congenital Disorder of Glycosylation type 1a
Congenital Fibrosis of the Extraocular Muscles
Congenital Myasthenic Syndrome (COLQ gene 603033) (Type Ic)
Congenital Secretory Chloride Diarrhoea
Congenital Stationary Night Blindness
Conradi-Hunermann-Happle Syndrome
Cowden syndrome (CS)/PTEN hamartoma tumour syndrome (PHTS)
Craniofrontal Dysplasia
Crouzon Syndrome
Cystic Fibrosis
Cystinosis
Czech dysplasia, metatarsal type also known as Progressive pseudorheumatoid dysplasia with hypoplastic toes
Dentatorubral-Pallidoluysian Atrophy (DRPLA)

Desbuquois Dysplasia (DBQD)
Diamond Blackfan Anaemia*
Diarrheoa 5 with tufting enteropathy congenital
Distal Hereditary Motor Neuropathy type IIB
Dominant Dystrophic Epidermolysis Bullosa
Donohue Syndrome
Downs syndrome
Dravet Syndrome
Dyskeratosis congenita (Male embryos only)
Dystonia 1 Torsion Autosomal Dominant (DYT1)
Early-onset Alzheimer disease Type 3 & 4
Ectodermal dysplasia (Hypohidrotic)
Ectrodactyly, Ectodermal Dysplasia, Clefting Syndrome (EEC)
Ehlers-Danlos Type IV
Elastin (ELN)-related Supravalvular Aortic Stenosis
Ellis-Van Crevald Syndrome
Epilepsy, female restricted, with mental retardation (EFMR)
Facioscapulohumeral Dystrophy
Factor XIII deficiency
Familial Adenomatous polyposis coli (FAP)
Familial Dysautonomia
Familial Hemophagocytic Lymphohistiocytosis (FHL)
Familial Hypertrophic Cardiomyopathy 4 (CMH4)
Familial Paranganglioma Syndrome (PGL1)
Fanconis Anaemia A*
Fanconis Anaemia C*
Fragile X Syndrome
Fraser Syndrome
Fried Syndrome
Frontotemporal Dementia
Galactosialidosis (early infantile and adult/ juvenile types)
Gangliosidosis (GM1)
Gaucher's Disease (Type II)
Gaucher Disease Type III
Glutaric Acidemia (aciduria)
Glycogen Storage Disease Type 1A
Gonadal mosaicism
Gorlin Syndrome
Greig's Cephalopolysyndactyly
Haemophilia A
Haemophilia B
Harlequin Ichthyosis
Hereditary diffuse gastric cancer
Hereditary Haemorrhagic Telangiectasia or Rendu-Osler-Weber Syndrome
Hereditary motor and sensory neuropathies
Hereditary Multiple Exostoses Type II

Hereditary Nonpolyposis Colorectal Cancer: Lynch Syndrome (for all subtypes)
Holt Oram Syndrome
Homozygous familial hypercholesterolaemia
Hunters Syndrome
Huntingtons Disease (Huntingtons Chorea)
Hydrocephalus
Hydroxyisobuyryl CoA Hydrolase Deficiency
Hyper-IgE Recurrent Infection Syndrome, Autosomal Dominant
Hyper IgM Syndrome - Hypogammaglobulinaemia*
Hypochondroplasia
Hypophosphatasia (Infantile/ Perinatal lethal)
Hypophosphatemic Rickets: X-linked dominant (Xlh)
Hypospadias (severe)
Ichthyosis
Idiopathic Arterial Calcification of Infancy
Incontinentia Pigmenti
Infantile Neuroaxonal Dystrophy 1
Inflammatory Bowel Disease, Early-onset (IBD28)
IPEX Syndrome (Immunodeficiency, Polyendocrinopathy and Enteropathy, X-Linked)
Juvenile Retinoschisis
Kearns Sayre Syndrome (KSS)/ Pearsons Marrow-Pancreas Syndrome (PMPS)
Krabbe Disease
L–2-Hydroxyglutaric aciduria
Leber's hereditary optic neuropathy / Lebers Optic atrophy
Leber Congenital Amaurosis
Leigh's (subacute necrotising encephalopathy of childhood)
Leigh Syndrome (Infantile Subacute Necrotising Encephalopathy)
Lenz syndrome
Lesch Nyan Syndrome
Lethal Multiple Pterygium Syndrome (LMPS)
Leukocyte Adhesion Deficiency (Type I)*
Li-Fraumeni Syndrome
Long Chain 3-hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)
Long QT Syndrome Types 1, 2, 3, 5 & 6
Lowe Oculocerebrorenal Syndrome
Lymphoproliferative Syndrome
Lynch syndrome / HNPCC (MLH1 gene)
Lynch syndrome / HNPCC (MSH2 gene)
Macular Dystrophy (childhood onset - variant of Retinitis pigmentosa)
Macular Dystrophy Retinal 2
Malignant Infantile Osteopetrosis
Maple Syrup Urine Disorder (MSUD)
Marfan Syndrome
Meckel-Gruber Syndrome Type 3

Medium-chain acyl-Co A dehydrogenase
Medium-Chain Acyl-CoA Dehydrogenase Deficiency
MELAS (Mitochondrial encephalomyopathy, lactic acidosis and stroke-like episodes)
Menkes Syndrome
Metachromatic Leukodystrophy
Methylmalonic Aciduria and Homocystinuria
Micro Syndrome (WARBM)
Mitochondrial DNA Depletion Syndrome 2 (myopathic type)
Mucolipidosis type II
Mucopolysaccharidosis III (MPS-III) Type B
Mucopolysaccharidosis III (MPS-III) Type C
Mucopolysaccharidosis III (MPS-III) Type D
Mucopolysaccharidosis Type I (MPS I)
Mucopolysaccharidosis type VI
Multiple acyl-CoA dehydrogenase deficiency (MADD) (also known as glutaric aciduria type II)
Multiple Endocrine Neoplasia (Type I)
Multiple Endocrine Neoplasia Type 2A (MEN type 2A)
Multiple Endocrine Neoplasia type 2B (MEN 2B)
Multiple Epiphyseal Dysplasia Type 5 (MED5)
Multiple Exostoses
Multiple Lentigines Syndrome (LEOPARD Syndrome)
Multiple Pterygium Syndrome Lethal Form(LMPS)
Muscle-Eye-Brain Disease
Muscular Dystrophy-dystroglycanopathy Type A5
Muscular Dystrophy (Beckers)
Muscular Dystrophy (Duchenne)
Muscular dystrophy (Occulopharangeal)
Muscular dystrophy, Limb-Girdle (LGMD) Type 1B
Myoclonic epilepsy and ragged red fibres (MERFF)
Myotonic Dystrophy
Myotublar myopathy
Nephrogenic Diabetes Insipidus (NDI)
Neurofibromatosis type I
Neurofibromatosis type II
Neurogenic muscle weakness, ataxia, retinitis pigmentosa (NARP)
Niemann Pick Disease Type A
Niemann Pick Disease Type C
Non-Ketotic Hyperglycinaemia (NKH)/ Glycine Encephalopathy (GCE)
Noonan Syndrome
Norrie Disease
Oculocutaneous Albinism Type 1A
Oculocutaneous Albinism Type 1B
Omenn Syndrome
Ornithine carbamoyl transferase Deficiency (OTC)
Ornithine transcarbamylase deficiency (OTD)

Osteogenesis Imperfecta (Type II)
Osteogenesis Imperfecta (Type III)
Osteogenesis Imperfecta type 1A
Osteogenesis Imperfecta type IV , type V , type VI
Osteogenesis Imperfecta Type1 (OI1)
Osteopetrosis with Renal Tubular Acidosis (OPTB3)
Osteopetrosis, Autosomal Recessive 5 and Osteopetrosis, Infantile Malignant 3
Ostheopathia Striata with Cranial Sclerosis (OSCS)
Otopalatodigital syndrome (Type 2)
Pachyonychia Congenita Type 1
Paragangliomas 4 (plg 4)
Partial Lipodystrophy, Familial (Type 2)
Pelizaeus Merzbacher Disease
Peroxisome Biogenesis Disorders PBD (Zellweger Syndrome Spectrum ZSS)
Phenylketonuria (PKU)
Plakophilin 1 (PKP1) associated ectodermal dysplasia syndrome
Polycystic kidney disease
Pompe Disease (early onset)
Pontocerebellar Hypoplasia type 1a, type 2a, type 2b, type 2c, type 2d , type 3, type 4, type 6
Pontocerebellar Hypoplasia type 1B (PCH1B)
Popliteal Pterigum Syndrome
Prader Willi Syndrome
Progressive Familial Intraheptic Chloestasis Cholestasis Type 1 (PFIC1)
Propionic Acidemia
Pseudoachondroplasia
Pseudohypoparathyroidism PHP1a
Pyrodoxine-dependent seizures
Pyruvate Dehydrogenase E1-beta Deficiency
Recessive Dystrophic Epidermolysis Bullosa* (Halleau-Siemens & Herlitz junctional)
Recurrent Digynic Triploidy
Recurrent hydatitiform mole
Renal Coloboma Syndrome
Retinitis Pigmentosa
Retinoblastoma
Retinoschisis (Juvenile)
Rett Syndrome RTT and Neonatal Encephalopathy
Rhesus disease/ Haemolytic Disease of the Newborn (HDN)
Rothmund-Thomson Syndrome (RTS)
Sandhoff Disease
Sanfilippo or Mucopolysaccharidosis Type III A
Sanjad Sakati Syndrome
Seathre-Chotzen
Senior Loken Syndrome 6

Sensorineural deafness - autosomal recessive non-syndromic
Severe Combined Immune Deficiency (x-linked)
Severe Combined Immunodeficiency – autosomal recessive
Severe Combined Immunodeficiency (SCID)
Severe Combined Immunodeficiency (SCID) (Adenosine Deaminase (ADA) deficient)
Sickle Cell Anaemia*
Simpson Golabi Behmel Syndrome Type 1
Smith Lemli Opitz Syndrome
Spastic paraplegia
Spinal and Bulbar Muscular Atrophy X-linked (Kennedy disease) (in affected males embryos)
Spinal Muscular Atrophy (SMA1)
Spinal Muscular Atrophy and Respiratory Distress (SMARD1)
Spinocerebellar Ataxia Type 1 (SCA1)
Spinocerebellar Ataxia Type 2 (SCA2)
Spinocerebellar Ataxia Type 3 (SCA 3) (Machado Joseph Disease)
Spinocerebellar Ataxia Type 6
Spondyloepiphyseal Dysplasia Congenita
Stickler Syndrome type 1, 2, 3 and autosomal recessive
Stuve-Wiedemann Syndrome
Succinic Semialdehyde Dehydrogenase Deficiency (SSADHD)
Surfactant Metabolism Dysfunction, Pulmonary 1 (SMDP1)
Tay Sachs Disease (infantile onset)
Torsion Dystonia
Townes-Brocks Syndrome
Treacher Collins Syndrome
Treacher Collins Syndrome Type 2 (TCS2)
Tuberous Sclerosis (TSC2)
Turner's syndrome (Mosaic)
Tyrosinaemia Type 1
Ullrich Muscular Dystrophy
Von Hippel Lindau (VHL) Syndrome
Walker Warburg Syndrome (Muscular dystrophy dystroglycanopathy)
Wiscott-Aldrich Syndrome*
Wolcott-Rallison Syndrome
Wolman's Disease (Acid Lipase Deficiency)
X-Linked Emery-Dreifuss Muscular Dystrophy (EDMD) (Male embryos only)
X-Linked Lymphoproliferative Disease Type 2 (XLP2) (Male Embryos Only)
X-Linked Thrombocytopenia (XLT)
X Linked Retinitis Pigmentosa (RP3)

* These conditions have also been licensed for use in cases involving HLA tissue typing. HLA tissue typing tests are licensed on a case-by-case basis, for specific patients.

The above list was taken from the HFEA website http://www.hfea.gov.uk on 26th March 2014.