

Red Cell Molecular Genotyping Information Sheet

Updated September 2017 Version 3.0

Red cell molecular genotyping is now a powerful tool in contemporary red cell antigen and antibody investigations. The Blood Service now offers red cell genotyping utilising the BioArray BeadChipTM Technology for a wide range of single nucleotide polymorphisms (SNPs) affecting antigen expression.

BeadChip Assay	Antigen / Variant Coverage	
HEA BeadChip	Rh Kell Duffy Kidd MNS Lutheran Dombrock Landsteiner Wiener Diego Colton Scianna	C/c, E/e, VS, V K/k, Js ^a /Js ^b , Kp ^a /Kp ^b Fy ^a /Fy ^b , Fy ^x , GATA Jk ^a /Jk ^b M/N/S/s, U-, Uvar Lu ^a /Lu ^b Do ^a /Do ^b , Hy+/Hy-, Jo(a+)/Jo(a-) Lw ^a /Lw ^b Di ^a /Di ^b Co ^a /Co ^b Sc1/Sc2
RHD BeadChip >75 RH variants	Weak D Type	1, 1.1, 2, 3, 4.0, 4.1, 4.2/DAR, 4.3, 5, 11, 14, 15, 17, 25, 29, 34, 40, 47, 51
	D negative	RHDψ; W16X; D-CE(3-7)-D; D-CE(4-7)-D; (C)dce°; D- CE(3-9); CE(1-3)-D(4-10); rG; RHD(Y269X)
	D _{el}	1227 G>A; IVS3+1G>A; M295I
	Partial D	DAR/weakD 4.2; DIIIa (DIII type 5), DIII type 4.6; DIIIc; DIVa; DIVa -2; DIV type 3,4,5; DIVb; DV type 1,2,3 (DBS-0), 4,5 (DHK), 6,7,8,9; DVI type 1,2,3,4; DNB;
RHCE BeadChip 48 RH variants	Wildtype	Ce, Ce, cE, CE
	ce variants	ce(48C), ce (48C, 697G, 733G, 1006T), ce(48C, 697G,712G, 733G, 916T), ce(48C, 733G, 744C)
	Partial ce	ceTI, ceAR, ceEK, ceMO, ce(733G), ce(48C, 733G), ce(48C, 733G, 1006T), ceCF, ce(697G, 733G), ceJAL, ce(48C, 340T, 733G),
	Partial e	ce(1025T), ce(48C, 733G, 1025T), ce(733G, 1006T),
	e+ ^w	ceSL, ceRT, ceRA, ce(48C, 733G, 748A), ceHAR,
	C+ ^{w/-} e+ ^w	CeMA, CeRN
	C+ e+	
	Partial E	CE(3031) CEEW CEEM CE(602C) CEKH
	E+ ^w	cE(344C)
		cE(365T)

The BioArray BeadChip uses the proprietary Elongation-mediated Multiplexed Analysis of Polymorphisms (eMAP) technology to identify the presence or absence of the selected alleles associated with a given phenotype.

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Clinical Indications

<u>HEA BeadChip</u> is recommended for the determination and prediction of **blood group** phenotypes in recently transfused patients or patients with autoimmune conditions where the patient's own immunoglobulins interfere with conventional grouping reagents. Unlike conventional phenotyping, BeadChip assays for red cell blood groups are not influenced by immunoglobulin coating of red blood cells, the presence of recently transfused red cells or any form of polyagglutination.

In addition, the Blood Service Red Cell Reference Laboratory Service may utilise this assay to assist with the **resolution of complex and challenging red cell antigen and antibody investigations** referred to us.

<u>RHD</u> BeadChip is used in conjunction with conventional serology to assist in determining the **precise RhD blood group status**, primarily in females who are pregnant, or who are of child bearing age.

<u>RHDCE BeadChip</u> is used in conjunction with conventional serology to assist in determining the **precise RhCE blood group status** in patients who are **suspected to have altered RhCE antigen expression**. The Blood Service Red Cell Reference Laboratory Service may perform this assay following results of the HEA assay or to assist in resolution of red cell antigen and antibody investigations suspected to be Rh related.

Amniotic fluid or cultured amniocytes can also be tested using any of the three assays to predict the fetal phenotype for determining the risk of Haemolytic Disease of the Foetus and Newborn (HDFN) in isoimmunised pregnancy.

Sample Requirements

- A minimum of 1 x 4mL EDTA whole blood (dedicated sample) is required. Lithium Heparin samples are NOT suitable.
- Or a minimum of 10mL of amniotic fluid or cultured amniocytes suspended in PBS. Cultured samples are preferred as they generally give a higher yield of DNA.
- Samples must be labelled with a minimum of 2 identifiers (prefer at least 3)
- Samples must be provided with completed request form with a minimum of 3 identifiers and include the tests requested, referring organisation's contact details and any relevant clinical information.
- The identifiers on the sample must match the request form exactly.

Note: Identifiers must include full name and at least one of either date of birth or unique medical record number.

Samples should be sent packaged with a cold ice-brick to the QLD Red Cell Reference Laboratory, via the address below as soon as possible following collection. Where there is a delay in transport (more than 12 hours from collection) the sample should be refrigerated.

QLD Red Cell Reference Laboratory Australian Red Cross Blood Service 44 Musk Avenue (delivery via Blamey Street) Kelvin Grove, Queensland AUSTRALIA 4059

Phone:+61 7 3838 9493 Fax: +61 7 3838 9410

Reporting

It is expected that results will be reported within 5 working days from receipt of the sample.

Enquiries

Please direct all enquiries to the Red Cell Reference Laboratory on +61 7 3838 9493.