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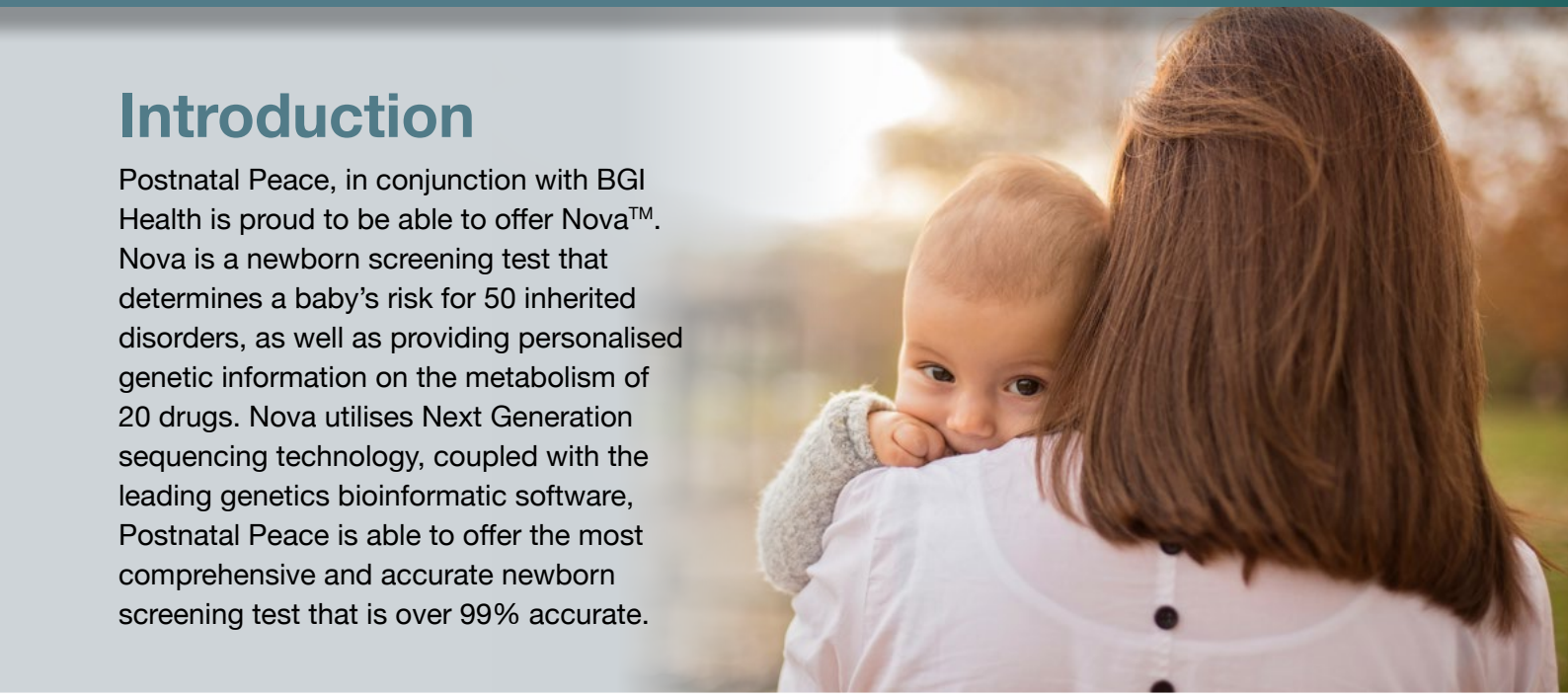
PERSONALISED MOLECULAR DIAGNOSTICS FOR INHERITED DISEASE FOR YOU

NOVA™ Newborn Genetic Test



Introduction

Postnatal Peace, in conjunction with BGI Health is proud to be able to offer Nova™. Nova is a newborn screening test that determines a baby's risk for 50 inherited disorders, as well as providing personalised genetic information on the metabolism of 20 drugs. Nova utilises Next Generation sequencing technology, coupled with the leading genetics bioinformatic software, Postnatal Peace is able to offer the most comprehensive and accurate newborn screening test that is over 99% accurate.

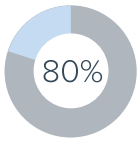
Why NOVA™ Newborn Genetic Testing?

Each year, over 7.9 million babies are born with birth defects, most of which appear perfectly healthy at birth and come from families with no history of the disorder. Many affected babies are not identified until the appearance of severe and often irreversible symptoms later in life.

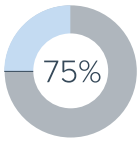
Many countries run publicly funded programmes to screen newborns for inherited disorders.

Currently, all Australian-born newborns are screened for five common conditions including congenital hypothyroidism, cystic fibrosis, amino acid disorders including phenylketonuria, organic acidemias and fatty acid oxidation defects. All states and territories except Victoria also screen for galactosaemia. There are a growing number of conditions that could be considered for inclusion in newborn screening programs and these include, among others, severe combined immunodeficiency and lysosomal storage disease (Newborn Screening in Australia: Position Statement May 2015, Royal Australasian College of Physicians). This leaves thousands of newborns unscreened for any number of potentially manageable disorders and adverse drug reactions.

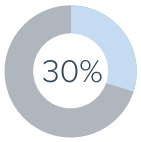
NOVA™ is suitable for newborns and children up to 5 years of age. NOVA™ screens for 50 inherited disorders, which have a combined prevalence rate of 1/400 births. NOVA™ aims to achieve early detection, referral and treatment of all babies identified as at high risk of these disorders.



of rare diseases have identified genetic origins*



of rare diseases affect children†



of rare disease patients die before the age of 5†

"1/17 will be affected by rare disease at some point in their life"‡

Sources: *eurodis.org, ‡European Council, †raredisease.org.uk

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What Does NOVA™ Test For?

50 Inherited Disorders (see table below)
Inherited Metabolic Diseases (40) / Congenital Hearing-impairment (1) / Immunodeficiency (6) / Other Monogenic Diseases (3)

20 Paediatric PharmacoPostnatal Peaces
Neurology Drugs (5) / Anti-infection Agents (10) / Rheumatology Drugs (1) / Gastroenterology Drugs (2) / Cardiology Drugs (1) / Oncology Drugs (1)

Condition Category	No.	Condition Name	Gene	Inherited Mode
Amino Acid Disorders	1	Phenylketonuria	PAH	AR
	2	Tetrahydrobiopterin(BH4)-deficient Hyperphenylalaninemia	FAH	AR
	3	Maple Syrup Urine Disease	BCKDHA	AR
			BCKDHB	AR
			DBT	AR
			DLD	AR
	4	Argininosuccinic Acidemia	ASL	AR
	5	Citrullinemia Type I	ASS1	AR
	6	Arginase Deficiency	ARG1	AR
	7	Carbamoylphosphate Synthetase I Deficiency	CPS1	AR
	8	N-Acetylglutamate Synthase Deficiency	NAGS	AR
	9	Ornithine Transcarbamylase Deficiency	OTC	XL
	10	Citrin Deficiency	SLC25A13	AR
	11	Homocystinuria Caused by Cystathionine Beta-Synthase Deficiency	CBS	AR
	12	Tyrosinemia Type 1	PTS	AR
Organic Acid Disorders	13	Methylmalonic Acidemia	MUT	AR
			MMAA	AR
			MMAB	AR
			MCEE	AR
			MMADHC	AR
	14	Propionic Acidemia	PCCA	AR
			PCCB	AR
	15	Isovaleric Acidemia	IVD	AR
	16	Carbamoylphosphate Synthetase I Deficiency	MCCC1	AR
			MCCC2	AR
	17	Glutaric Acidemia Type I	GCDH	AR
	18	Beta-Ketothiolase Deficiency	ACAT1	AR
	19	Beta-Ketothiolase Deficiency	BTD	AR
				AR
			HLCS	AR
Fatty Acid Oxidation Disorders	20	Glutaric Acidemia type II	ETFDH	AR
			ETFA	AR
			ETFB	AR
	21	Systemic Primary Carnitine Deficiency	SLC22A5	AR
	22	Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	HADHA	AR
	23	Medium-Chain Acyl-Coenzyme A Dehydrogenase Deficiency	ACADM	AR
	24	Trifunctional Protein Deficiency	HADHA	AR
			HADHB	AR

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Condition Category	No.	Condition Name	Gene	Inherited Mode
Fatty Acid Oxidation Disorders	25	Very Long-Chain Acyl-Coenzyme A Dehydrogenase Deficiency	ACADVL	AR
	26	Carnitine Palmitoyltransferase II Deficiency	CPT2	AR
	27	Carnitine Palmitoyltransferase 1A Deficiency	CPT1A	AR
	28	Short-Chain Acyl-CoA Dehydrogenase Deficiency	ACADS	AR
Copper Metabolism Disorder	29	Wilson Disease	ACADS	AR
Carbohydrate Disorders	30	Glucose-6-Phosphate Dehydrogenase Deficiency	G6PD	XL
	31	Hereditary Fructose Intolerance	ALDOB	AR
	32	Galactosemia	GALT	AR
			GALE	AR
			GALK1	AR
Lysosomal Storage Diseases	33	Fabry Disease	GLA	XR
	34	Glycogen Storage Disease Type Ia	G6PC	AR
	35	Glycogen Storage Disease Type Ib	SLC37A4	AR
	36	Glycogen Storage Disease Type II (Pompe Disease)	GAA	AR
	37	Mucopolysaccharidosis Type I	IDUA	AR
	38	Mucopolysaccharidosis Type II	DLD	XR
	39	Krabbe Disease	GALC	AR
	40	Niemann-Pick Disease	SMPD1	AR
			NPC1	AR
			NPC2	AR
Hearing Impairment	41	Nonsyndromic Hearing Loss and Deafness	GJB2	AD/AR
			SLC26A4	AR
			GJB3	AD/AR
			MT-RNR1	Mitochondrial Inheritance
Primary Immunological Deficiency	42	Severe Combined Immunodificiency	IL2RG	XR
			JAK3	AR
			IL7R	AR
			PTPRC	AR
			CD3D	AR
			CD3E	AR
			CD247	AR
			RAG1	AR
			RAG2	AR
			DCLRE1C	AR
			AK2	AR
			ADA	AR
			LIG4	AR
			NHEJ1	AR
			PNP	AR
			ZAP70	AR
	43	Beta-Ketothiolase Deficiency	BTK	XR
	44	Ataxia-Telangiectasia	ATM	AR
	45	Nijmegen Breakage Syndrome	NBN	AR
	46	Cartilage Hair Hypoplasia	RMRP	AR
	47	Familial Hemophagocytic Lymphohistiocytosis	PRF1	AR
			UNC13D	AR
			STX11	AR
			STXBP2	AR
Miscellaneous Genetic Conditions	48	Cystic Fibrosis	CFTR	AR
	49	Severe Myoclonic Epilepsy of Infancy	SCN1A	AD
	50	Tuberous Sclerosis	TSC1	AD
			TSC2	AD

The NOVA™ Newborn Genetic Test screens for mutations which have been linked to the specific genetic conditions listed on the testing panel. The purpose of the NOVA™ Newborn Genetic Test is to identify babies as more likely to have one of the listed genetic conditions. If the test result returns as positive for one of the mutations, definitive diagnosis of the condition should only be undertaken by a qualified healthcare professional. Further, confirmatory diagnostic testing is recommended.

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Who is NOVA™ Suitable for?

NOVA™ is particularly suitable for:

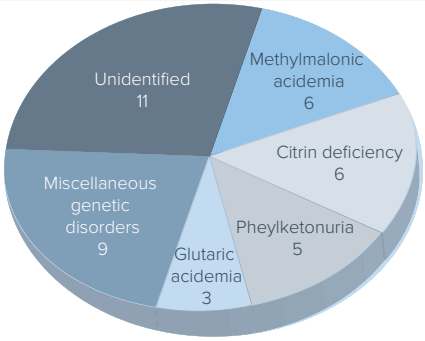
- Parents who want a comprehensive genetic screen for their baby or child up to 5 years old.
- Parents who would like to learn their baby's drug-related genetic status
- Babies who have missed out on regular screening
- Babies from parents with a family history of inherited disorders or from a population identified as at higher risk for genetic disease
- Parents who have suffered habitual abortion, stillbirth or neonatal mortality

NOVA™ is NOT suitable for:

- Definitive diagnosis of a disorder
- Newborns with numerical or structural changes of the chromosome, copy number variations and/or germ cell mosaicism
- Newborns who have received blood transfusions, organ transplants or stem cell therapy

Clinical Validation:

40 samples



Gene mutations consistent with clinical phenotypes were discovered in 29 (72.5%) samples. The other samples were only discovered to have common pathogenic mutations or SNPs.

Rare Disease Information Resources

"Even those with severe rare diseases can sometimes be identified and treated at an early stage to reduce the impact of their disease (for example through surgery, nutrition or medication). Antenatal and new born screening (for example newborn blood spot screening) has an important role to play." *Source: UK Strategy For Rare Diseases, p14*

"Rare diseases are characterised by a wide diversity of symptoms and signs that vary not only from disease to disease but also from patient to patient suffering from the same disease. Relatively common symptoms can hide underlying rare diseases, leading to misdiagnosis." *Source: www.eurordis.org/content/what-rare-disease*

"A rare disease is defined by the European Union as one that affects less than 5 in 10,000 of the general population. There are between 6,000 and 8,000 known rare diseases. Around five new rare diseases are described in the medical literature each week." *Source: www.raredisease.org.uk/about-rare-diseases*

Publications

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Thong MK. *Birth defects registries in the Postnatal Peaces era: challenges and opportunities for developing countries.* Front Pediatr. 2014 Jun 16;2:60. doi: 10.3389/fped.2014.00060. eCollection 2014.

Rieder MJ, Carleton B. *PharmacoPostnatal Peaces and adverse drug reactions in children.* Front Genet. 2014 Apr 16;5:78. doi: 10.3389/fgene.2014.00078. eCollection 2014.

Brandi ML. *Lessons from next-generation sequencing in genetic skeletal disorders.* Bonekey Rep. 2014 May 14;3:528. doi: 10.1038/bonekey.2014.23. eCollection 2014.

Waisbren SE. et.al.*Parents are interested in newborn Postnatal Peace testing during the early postpartum period.* Genet Med. 2014 Dec 4. doi: 10.1038/gim.2014.139.

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