

Customized kits supporting diagnosis of genetic disorders

Clinical sample collection packs

Solution	
Product	Trajan custom Children's Hospital at Westmead (CHW) cerebrospinal fluid (CSF) clinical sample collection pack
Sample	Cerebrospinal fluid (CSF)
Site	Children's Hospital at Westmead (CHW), New South Wales, Australia.

Introduction

Trajan Scientific and Medical (Trajan) is pleased to support genetic diagnostic services via the Children's Hospital at Westmead (CHW), New South Wales, Australia.

CHW is part of the Sydney Children's Hospital Network (SCHN), the largest network of hospital and services for children in Australia.

Their diagnostic and support services cover not only Sydney, but a large area of the Asia Pacific region, for example, accepting cases from Australia, New Zealand, Malaysia and Singapore.



Figure 1. Analyzing the cerebrospinal fluid (CSF) specimen using UPLC in the laboratory. Shirley Alvarez, Hospital Scientist, the Children's Hospital at Westmead.



Figure 2. Example cerebrospinal fluid clinical sample collection pack

Summary

Trajan has worked closely with CHW Neurochemistry and Biochemistry laboratories, to develop a customized kit to improve cerebrospinal fluid (CSF) sample collection during newborn screening for nonketotic hyperglycinaemia (NKH).

NKH is a genetic disorder that interferes with cellular metabolic processes. Early diagnosis and intervention is critical as NKH can be fatal, or affect many different organs causing severe problems with learning and development.

Within Trajan's customized CHW CSF Sampling Kit, CSF tubes are labelled to identify the required volume and indicate the specific test, and include an amber tube to ensure reduced light exposure to help maintain sample integrity.

Dr Sushil Bandodkar, Principal Scientist and Head, Biochemistry, CHW, said, "Having the customized CSF collection kits ready to go saves our team time from the collection stage through to analyzing the specimen in the laboratory."

"The prepared kits allow improved sample integrity."

The CSF kits are made up in Trajan's clinical diagnostic packing facility based in Victoria, Australia. This is a clean monitored temperature-controlled environment, purpose-built to meet the needs of healthcare and pathology providers through the supply of TGA compliant customized diagnostic procedure packs or kits intended for our customer's in-house use.

By working with Trajan, laboratories can have specialized kits made up to required standard, saving administration time and money. Trajan supplies customized packs as per the customer's specification.

SCHN and CHW

Sydney Children's Hospitals Network

The Sydney Children's Hospitals Network (SCHN) incorporates The Children's Hospital at Westmead, Sydney Children's Hospital, Randwick, Bear Cottage, the Newborn and pediatric Emergency Transport Service (NETS), the Pregnancy and newborn Services Network (PSN) and the Children's Court Clinic.

SCHN is the largest network of hospital and services for children in Australia. Each year the network services manage 51,000 inpatient admissions, 92,000 Emergency Department presentations, and over one million outpatient service visits.

Children's Hospital at Westmead Pediatric Pathology Services – Biochemistry and neurochemistry

The Children's Hospital at Westmead's (CHW) Biochemistry laboratory offer a comprehensive range of biochemistry services, including:

- Galactosaemia testing
- Urinary PHPAA and histamine testing
- Urinary pterin and biogenic amine testing
- Sweat tests

The CHW Australasian Paediatric Neurochemistry Laboratory is a highly specialized unit within Biochemistry which measures neurotransmitters, their metabolites, precursors and pteridine cofactors in cerebrospinal fluid and urine and receives samples from all states across Australia and nearby countries such as New Zealand, Singapore, and Malaysia.

These tests detect neurodegenerative, inflammatory central nervous system (CNS) diseases and movement disorders affecting the brain and



Figure 3. Alex Anderson, Trajan Scientific and Medical with Dr Sushil Bandodkar, Principal Scientist and Head, Biochemistry, The Children's Hospital at Westmead, New South Wales, Australia.

spinal cord and inborn-errors of neurotransmitter biosynthesis and metabolism including defects or deficiencies in pteridine biochemistry (including DOPA responsive Dystonia, and Dystonia/ Myoclonus Syndrome); tyrosine hydroxylase, aromatic amino acid decarboxylase, dopamine-beta-hydroxylase, monoamine oxidase and catechol-o-methyltransferase deficiencies.

The unit analyses dopamine and metabolites for detecting neuroblastoma and ganglioneuroma, noradrenaline, adrenaline and metabolites for detecting pheochromocytoma and histamine for systemic mastocytosis. Special loading tests such as Phenylalanine and/or Tetrahydropterin challenge testing is also conducted in the Neurochemistry Laboratory for the differential diagnosis of Pterin deficiencies and classical PKU.

Conclusion

Trajan custom clinical sample collection packs

- Custom packs provide access to the right products required for the procedure type in one pack, thereby reducing inaccuracies and promote efficiency in front-line patient care with better time management.

Acknowledgments: Dr Sushil Bandodkar, Principal Scientist and Head, Biochemistry, and Shirley Alvarez, Hospital Scientist, The Children's Hospital at Westmead, New South Wales, Australia.

Information and support

Visit www.trajanscimed.com or contact techsupport@trajanscimed.com

Specifications are subject to change without notice.