



Investigations for Children with Significant Developmental Delay

By Dr Stephen Withers

A child with a global developmental delay places a major social and emotional burden on a family. The range of possible causes is wide but if a firm diagnosis can be reached, discussion of prognosis and future expectations becomes possible. This applies not only to the current child but also to other siblings. Considerations of treatment (and several diseases are eminently amenable to treatment once diagnosed) and counselling before future pregnancies can be considered.

A systematic approach to investigation is needed to most effectively reach a diagnosis. Even if a referring doctor is not making a full evaluation, having some of these results available at the time of referral to a specialist or sub-specialist can be valuable in narrowing down further studies.

Initial Testing

Along with detailed history and examination, initial testing with full blood count, electrolytes and liver function tests, and urine metabolic screen is recommended. The urine metabolic screen is useful because the neonatal screen currently only tests for a limited set of metabolic disorders. If a neonatal screen has been performed and results identified, then thyroid function screening will have been performed, but a need for repeat thyroid testing should be determined on clinical features. If muscle weakness is present, serum creatine kinase (CK) should be measured.

Family History

At times, history will identify a close family member with global developmental delay. If so, and if the

nature of the disorder is known then directed testing is indicated. Otherwise, with close family history, cytogenetic analysis (karyotype and Fragile X testing) should be requested.

Fragile X is the most common recognised cause of significant developmental disability in males. It must be remembered however that females may both carry and be affected by this condition. Testing is available through QML.

Progressive advances in technology have allowed us to look at the chromosomes closely, particularly looking for small deletions or rearrangements. This may be important for the child affected by the condition, but it may bear important implications for the parents and for prospects of future pregnancies should one of them be found to carry a balanced chromosomal ("cryptic") translocation.

Specific Features

In the absence of a positive family history, further testing plans are best guided by features suggesting a specific diagnosis. Features of Down Syndrome, Fragile X, hypothyroidism, or dysmorphic appearance should be sought and specific testing requested if present.

If clinical evaluation reveals microcephaly, history of intrapartum stress, or focal features that may suggest CNS injury or malformation, then CNS imaging should be discussed with an imaging specialist.

If identifiable risk factors for excessive environmental lead exposure at above established guidelines are present then blood lead analysis should be undertaken.

continued inside...

Warfarin reminder

**PLEASE REMEMBER to register your new Warfarin Care Patients on 1300 795 355
BEFORE they present for testing**

- > **NEW PATIENTS ONLY** need to be registered
- > **If you need a new info pack/sticker** please contact QML Liaison on (07) 3840 4539
- > **Please be patient on the 1300 line** – we are adding extra staff to deal with enquiries and registration

CPD Seminar Program

AMA Health and Lifestyle Expo

Saturday 24 July 2004
Brisbane Convention Centre, South Brisbane

AMAQ in conjunction with QML Pathology are once again proud to present a series of Continuing Professional Development seminars as part of the AMA Health and Lifestyle Expo.

Designed specifically for medical practitioners, these one hour CPD's* give you the flexibility of attending a single seminar or a comprehensive set of four. *2 CPD points/hr (subject to RACGP approval).

This year's AMA Health and Lifestyle Expo CPDs concentrate on the broad base of Tumors and their effects, with an introductory discussion on Tumor markers and their uses for Oncology patients. A further investigation into classifications prevalence and investigations of colorectal CA will be covered. Prostatic CA and PSA will also be discussed in depth with a focus on incidence, prognosis and testing. The last lecture will conclude with new Oncology today, new treatments and palliative care for General Practice.

Family Pass

AMA Health and Lifestyle Expo



Admit 2 adults and 3 children
July 23-25 from 10am-5pm
Brisbane Convention Centre

www.amahealthexpo.com.au



CPD Program for Saturday, 24 July

(please ✓ below to indicate which sessions you wish to attend)

- 9.30am-10.30am **Serum Tumor Markers**, Speakers: Dr Kerry DeVoss
- 11.00am-12.00pm **Urology Issues: Prostate and PSA**, Speakers: Dr John Preston
- 1.00pm-2.00pm **Colorectal Issues**, Speakers: Dr Michael Mar Fan
- 2.30pm-3.30pm **Oncology Today**, Speakers: Dr Rick Abraham

Complimentary lunch, morning/afternoon tea & wine tasting session for all delegates.

Reserve Your Seat

PLEASE FAX THIS FORM BACK TO QML PATHOLOGY ON (07) 3840 4972
OR RETURN TO QML MARKETING VIA YOUR QML COURIER.

- Yes, I will be attending the session/s indicated (please tick session times above)
- No, I will not be attending

General Practitioner

Surgery

For Further Information, Please Contact QML Pathology On (07) 3840 4506.

Proudly sponsored by



Upcoming Events...

QML Southport Grand Rounds for 2004

Venue: QML Conference Room, Pindara Pathology Centre, Carrara Street Entrance (behind Pindara Kindyland)

Time: 7.00am – 8.00am (A light Continental style breakfast will be served)

Date: 7 July 2004

Topic: Non-Interventional Cardiology

Speaker: Dr Bryan Farage

Date: 4 August 2004

Topic: Interventional Gastroenterology

Speaker: Dr Galib Ali

Date: 1 September 2004

Topic: Rheumatology

Speaker: Dr Julien De Jager

Date: 6 October 2004

Topic: Interventional Cardiology

Speaker: Dr Yahya Kiwan

Date: 3 November 2004

Topic: Dermatology

Speaker: Dr Michael Freeman

Date: 1 December 2004

Topic: A Mixed Bag – Prize for best Presentation

Speaker: All comers – attendees to vote

St Andrew's CPD Program

St Andrew's Hospital hosts a comprehensive range of medical education events throughout the year. Below is a list of upcoming CPDs to be held throughout South East Queensland.

As numbers are limited, we encourage you to reserve your place - register your interest by calling (07) 3834 4293, fax (07) 3834 4576 or e-mail jscott@sawmh.com.au.

St Andrew's Orthopaedic CPD Weekend

Date: 7 – 8 August

Venue: Sheraton Resort, Noosa

Focusing on a range of topics specific to Orthopaedic Surgery and related fields in General Practice, the educational component of this weekend will centre on patient presentations with disorders of the major joints.

This meeting is designed to give GPs an update on Orthopaedic Patient Presentation through hands-on examination and assessment techniques. Several Orthopaedic Surgeons will present on each disorder and then break into small groups for more personalised learning.

St Andrew's Heart Institute CPD Weekend

Date: 4 – 5 September

Venue: Palazzo Versace, Gold Coast

This weekend will focus on a range of topics specific to Cardiology, Cardiac Surgery and Vascular Surgery in General Practice.

St Andrew's CPD Weekend

Date: 30 –31 October

Venue: Radisson Resort, Gold Coast

This CPD weekend will focus on managing different patient presentations in General Practice.

... from front page

Cases Without Specific Features

In the absence of recognised specific indications and if there is loss or regression of developmental milestones, then comprehensive investigation with imaging, metabolic and cytogenetic screening, and specialist genetics consultation is recommended. In other cases, stepwise evaluation of CNS imaging, cytogenetic screen / Fragile X testing and metabolic screening can be undertaken.

Many of these investigations need only be performed once and provide a sound platform for further clinical consideration. However the pursuit and establishment of a diagnosis is a worthwhile exercise because it can provide the family with advantages

of specific counselling as well as access to both services and financial assistance through state and federal agencies.

QML consultants are available to discuss planning and interpretation of these investigations. Please contact your local branch laboratory or QML Brisbane Medical Liaison on (07) 3840 4539.



Dr Stephen Withers

Dr. Stephen Withers has recently joined the QML Team to provide a Clinical Genetics Consultancy with respect to genetic testing and genetic results.

Dr. Stephen Withers is a graduate of the University of Queensland and completed training in Paediatrics at the Royal Children's Hospital. Dr. Withers subsequently embarked on training in clinical genetics working with the Queensland Clinical Genetic Service and completed training at the Sydney Children's Hospital.

He then undertook a post fellowship training year in Cancer Genetics with the Institute of Oncology at Prince of Wales Hospital. In 1999 Dr. Withers travelled to Canada and worked at the Hospital for Sick Children in Toronto. He returned to Australia in mid 2000 as the Director of Paediatrics at Logan Hospital. In 2004 he has taken up a post with the new Griffith Medical School based at the Gold Coast and will be working with QML to enhance and value add to the wide variety of genetics services which are currently available.

Dr. Withers can be contacted through QML regarding special testing or interpretation of genetic results. Please contact the QML Genetics Department for further information on (07) 3840 4461.



COLLECTION CENTRE NEWS

For the convenience of our doctors and patients, we have listed the latest additions and changes to the QML network of clinics:

NEW CLINIC

MAROOCHYDORE (07) 5479 5344

31-33 Plaza Parade, Maroochydore

Mon – Fri 8:00am – 4:00pm
(closed for lunch 12-12:30pm)

COLLECTION CENTRE CHANGES

LISMORE

(new opening hours) (02) 6621 4488

69 Uralba St

Mon-Fri 8:00am – 1:00pm

MACKAY

(new opening hours) (07) 4951 2999

29A Brisbane St

Mon-Fri 8:00am – 5:00pm

Sat 8:30am – 12 noon

FLECKER HOUSE

(new opening hours) (07) 4051 6961

4th Floor, Flecker House, Upward St

Mon-Fri 8:00am – 4:00pm
(closed for lunch 12-12:30pm)

MISSION BEACH

(new opening hours) (07) 4068 9175

Lot 11, Cassowary Dr, Wongaling Beach

Mon-Fri 8:00am – 5:00pm
(closed for lunch 12:30pm – 1:30pm)

Sat 8:00am – 11:30am

BYRON BAY

(new opening hours) (02) 6685 6681

52 Shirley St

Mon-Fri 8:30am – 5:00pm
(closed for lunch 1-2pm)

Please contact your local branch or Brisbane Liaison for further information on (07) 3840 4539.

QML Cardiac Pack

QML Pathology has recently created a Cardiac Information Pack for all GPs and Specialists. The pack outlines the following topics:

- Cardiac Troponin
- B-type Natriuretic Peptide
- Homocysteine
- High Sensitivity C-Reactive Protein
- Warfarin Information
- Thrombophilia

- Anticoagulation
- Prosthetic Heart Valves

The QML Cardiac Pack also contains comprehensive reference materials from the Australian Heart Foundation and the National Prescribing Service.

Please contact your local Medical Liaison Officer, or Brisbane Liaison on (07) 3840 4539 to order a complimentary copy of the pack.



Mayne Health Pathology Pty Ltd ABN 84 007 190 043 t/a Queensland Medical Laboratory

This newsletter has been prepared and published by QML Pathology for the information of referring doctors. Although every effort has been made to ensure that the newsletter is free from error or omission, readers are advised that the newsletter is not a substitute for detailed professional advice.

© Copyright June 2004



MAR/PUB/001 June 2004