香港兒童腦科及體智發展學會

HKCNDP

The Hong Kong Society of Child Neurology
& Developmental Paediatrics

Annual Scientific Meeting

8 - 11 March, 2002
Hong Kong

Paediatric Neuro-Ophthalmology

Course Director

Dr. David S.I. Taylor
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Great Ormond Street Hospital for Children, NHS Trust &
Visual Sciences Unit, Institute of Child Health
Great Ormond Street
London

The Hong Kong Society of Child Neurology & Developmental Paediatrics

2002 Annual Scientific Meeting
8 - 11 March 2002

Scientific Program
Topic : "Paediatric Neuro-Ophthalmology"

	8 Mar, (Friday)	9 Mar, (Saturday)	10 Mar, (Sunday)	11 Mar, (Monday)
Venue	QEH Blk M G/F	QEH Blk M G/F	QEH Blk M G/F	Great Eagle Hotel
AM			**Seminar III** 9:30-10:30 "Peculiar Visual Images" - Dr. David Taylor 10:30-11:15 Discussion & Tea 11:15-12:15 Local Presentation 1. "Developmental Management of Severe Visual Impairment- Experience of Child Assessment Service" - Dr. Iris Lau 2. "Assessment and Management of Strabismus and Amblyopia in Children" - Ms. Frenchy Chiu 12:15-12:30 Discussion	
Lunch		12:30 - 14:00	12:30 - 14:00	
PM		**Seminar II** 14:00–15:00 Local Presentation 1. "Acquired VI n Palsy in Children - a benign case and a not-so-benign case" - Dr. C.Y. Ko 2. "Paediatric Ophthalmic Assessment for Children with Severe, Multiple Disabilities" - Dr. C.H. Ko 15:00- 15:30 Discussion & Tea 15:30-16:30 " Eye Movement Disorders and Strabismus Syndromes" – Dr. David Taylor 16:30- 17:00 Discussion	**Seminar IV** 14:00–15:30 Free Paper Session 15:30- 16:00 Tea 16:00- 17:00 "Retinal Disease-When do I call the Neurologists?" - Dr. David Taylor	
Evening	**Seminar I** 19:00 –20:00 Light Buffet 20:00-21:00 "Optic Nerve and the Brain" - Dr. David Taylor 21:00-21:15 Discussion			**Plenary Lecture** 19:00 Cocktail 20:00-21:00 "The Apparently Blind Child" - Dr. David Taylor 21:00-22:00 Chinese Banquet Dinner

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Organizing Committee:

Dr. Chan Chok Wan
Dr. Catherine Lam Chi Chin
Dr. Philomena Tse
Dr. Winnie Yam Ka Ling
Dr. Tsui Kwing Wan
Dr. Wu Shun Ping
Ms Anciently Chan, Wyeth (HK) Ltd.

Course Director

Résumé CV of DAVID TAYLOR

DOB 6.6.'42, Hobart, Australia
Senior Lecturer
Institute of Child Health
Consultant Ophthalmologist
Great Ormond Street Hospital for Children
London

PROFESSIONAL QUALIFICATIONS:

FRCS	1973	FRCPCH 1997
FRCP	1984	D Sc (Med) 2001
FRCOphth	1990	

RESEARCH GRANTS
£ 4.7million in research grants and capital funds

MEMBERSHIP OF EDITORIAL BOARDS:
British Journal of Ophthalmology 1977-90 and 2000-
European Journal of Ophthalmology 1998-

NAMED LECTURES, MEDALS, AWARDS:
13, including:-

The Treacher Collins Prize OSUK	1982
The Richardson Cross Memorial Lecture and Medal	1991
The Doyne Lecture and Medal (Oxford)	1997
The Mackenzie Memorial Lecture and Medal (Glasgow)	1999
The Montgomery Lecture and Medal (Dublin)	1999

TEACHING AND RELATED ACTIVITY:
Children's Eye Group (Founder & Treasurer)
Paediatric Ophthalmology Courses (Annually with Inst. of Ophthalmol, jointly,)
British Council Course UK, biannually (+6 x Overseas)
Paediatric Ophthalmology and Strabismus in the 21st Century meeting Jan 2001
Founder and treasurer CEG/ HACTS traveling fellowship & Claud Worth Visiting Professorship

OVERSEAS INVITED LECTURES, PROFESSORSHIPSetc.
70 Overseas lectures and 5 visiting Professorships since 1985

PUBLICATIONS--224

Seminars

The Optic Nerve and the Brain - Development and Disorders

David Taylor, London

Symptoms in optic nerve and Chiasmal Disease

Visual
Headaches
Precocious or delayed puberty
Growth Disturbance
Sleep Disturbance
Temperature irregularity

Clinical Investigation

–VA
–Colour Vision
–Fields
Direct Ophthalmoscopy
–90D

Fundoscopy

-optic atrophy
-Bow-tie atrophy
-twin peaks papilloedema

Pupils

Further Tests

Neurophysiology
Plain X-rays
CT/MRI
Endocrine, sleep etc

DeVile C.J. et. al.1997
Timms C. et al. 1998

75 patients, 20(27%) H/O Squint.16 had squint
9(45%) of these were concomitant, 8/9 had optic atrophy at presentation

New Axons grow straight towards fetal fissure, all exit "Error Free"
Early retinal axon trajectories correlate with expression of regulatory genes: -
BF-1 BF2 Dlx-2 EphB2/3, Nkxx-2.1 Nkxx-2.2 Shh
Nerve,Chiasm & tract GAP 43 (Netrin1) &DCC Hesx1

Seminars

ONH- Inheritance?

Familial Cases

Missiroli 1947, Kytila & Miettinen 1961, Hachenbruch et al.1975, Benner et al. 1980, Wales & Quarrell 1996, Dattani 1998

BUT! Low Recurrence Risk

Achiasmia

–Midline defects, See-saw Nystagmus, Developmental Delay, VEP Defects , disc anomalies and ONH

Apkarian et. al. Eur J Neurosci 1994;6:501-7

Thompson D. et. al. Ophthalmology 1999;1062354-61

Chiasmal Neuritis

Newman NJ et al Neurology 1991; 41 : 1203-10

Scott IU et al AJO 1997 ; 123 : 136-138

Pomeranz & Lessell Arch Ophthalmol. 1999; 117: 128-131

Topless Optic Discs"Superior Segmental Hypoplasia"

- 34 Children of 23 diabetic mothers, 8% had topless discs
  Landau et al.AJO1998;125:605-11
- 4Cases of topless discs . Mother normal
  Hashimoto et al. AJO1999;128:111-112

ONH-why do thet die?

- Associated Brain Defects
- Because they're Blind
- Endocrine defects
- –Birth
- – Later Post. Pit. defects
- Shock Cascade

Seminars

ONH with Endocrine disturbances

- M>F

Post Pit ectop.=ant pit def.
(Brodsky&Glasier'93, Brodsky'99)
ONH+ no post pit = ant & post. Pit. Deficiency
(Sorkin et al.1999)
gonadotrophin preferentially spared
Nanduri& Stanhope 1999

Optic Glioma Regression

Parsa et al Arch. Ophthalmol 2001;119:516-529
13 pts, biopsy or radiology proven. 11 MRI, 2 CT serial changes in signal & size12/13 spontaneous tumour shrinkage-mostly marked
1/13 signal change without size change
At least 6 other papers

ONH and Tumours

- Keane 1977
- Osher& Schatz 1979
- Taylor 1982. ONG + Cranios
- Lee et al. 1997. Teratoma

Pseudochiasmal Disease

Coloboma Phenotypes

Coloboma-common Syndrome associations

Coloboma with cyst

Midline facial and brain disease and the optic disc

ON morphology that reflects adverse prenatal events

Optic Nerve Aplasia

Seminars

Strabismus Syndromes and Saccades

David Taylor, London

Strabismus Syndromes

1). Möbius Syndrome
- and other congenital Mobius-like Syndromes
- Horizontal gaze palsy +VIN palsy
- V, VII + other cranial nerve palsies
- Systemic Anomalies
- Inheritance
- Aetiology
- Social effects

2). Generalised Ocular Fibrosis Syndrome
- "convergence only" defect
- Is it a brainstem aplasia, too?

3). Duanes Syndrome
- Typical/atypical
- Up or down shoots
- Management
- Aetiology
  - Brainstem aplasia- embryopathy
  - Fibrosis
- Associated diseases
  - Various!
  - Wilderwancks syndrome

4). Brown's Syndrome
- clinical features
- management

5). Double Elevator Palsy
- clinical features
- management

Seminars

Saccades and their disorders in children-
VHS PAL Video

1). What do saccades do?

2). How do we measure saccades?

3). Development of saccades

4). Abnormal Saccades

a. Saccade initiation failure(COMA)

b. Slow saccades

c. Fast saccades

d. Intrusive saccades
   1. Square wave jerks
   2. Flutter-like oscillations
   3. Opsoclonus
   4. Macrosaccadic oscillations

e. Disorders of saccadic accuracy
   1. Hypometria
   2. Hypermetria

f. Neural Integrator failure

g. Voluntary Nystagmus

Peculiar Visual Images

David Taylor

Peculiar =
"nlike others, singular,strange,
odd, queer"
OED

Size Changes
(Dysmetropsia)

- Central
- Peripheral
- AC/A unlinking

Mis-interpretation of Normal Phenomena

- Physiological Diplopia
- Floaters
- Troxler's Phenomenon
- Scheerer's Phenomenon

Colour Changes

- Afterimages
- Coloured Clouds
- Interocular adaptation difference
- Erythropsia

Mis-interpretation of Normal Phenomena

- Massage Phosphenes
- Eye Movement Phosphenes
- Retinal Vessel Viewing by light stimulation
- Moore's Lightening Streaks

Distortions
(Metamorphopsia)

- Simple
- Intrusions

Eye Movement Phosphenes

- Seen in dark adapted eyes
- Nasal or temporal field
- Rapid movement
- muscle insertion
- Vitreous deformation
- Optic nerve stimulation

Nebel Arch. Ophthalmol 1957;58:235-243

Visual Perseveration

Perseveration=Persistence of an object in the absence of it's original external stimulus

In Time-Palinopsia

In Space-Polyopia

Moore's Lightening streaks

- F>M in middle age
- Temporal Field
- Last seconds
- Vertical, flashing
- DD=Retinal detachment

Foster Moore BJO 1935;19: 545-547

Palinopsia

"Visual Perseveration in Time" (Critchley)

'Vctive' occipital lobe lesions
Usually non-dominant Hemisphere

Usually associated Hemianopia
May have Hallucinations as well

Hallucinations

- Psychiatric Disease
- Localised Brain Disease
- Normal
  - -Hypnopompic
  - -Hypnagogic

Visual Phenomena in Blind Eyes

- Charles Bonnet's Syndrome
- Local phenomena

Hallucinations

- Peduncular Hallucinosis
- Social Deprivation
- Psychoneuroses and Behaviour Problems (Pseudohallucinations)
- Occipital Epilepsy

Visual Phenomena in the Blind Charles Bonnet's Syndrome

- Bilateral Blindness
- Hallucinations without delusions or loss of insightful cognition
- Vivid, formed, hallucinations
- Hallucinations usually lack a personal meaning, not frightening

White & Jan Dev. Med.Ch. Neurol.1992; 34:252-265

Peduncular Hallucinosis

- Midbrain or Thalamic Lesions
- Vivid, Terrifying Hallucinations
- Gaze Palsy

Visual phenomena in blind eyes *Unilateral blindness*

- Photopsias or Unformed Images
- Only when other eye closed
- "Builds up"
- May become semi complex or locally complex

Occipital Epilepsy

- Elementary visual Hallucinations
- multiple,bright, coloured spots,circles or balls
- Last for **seconds**
- Start temporally, move across the field
- Flashing

Auditory-evoked Phosphenes

- Optic Neuropathy+loud, unexpected sound
- --> Brilliant Flashes in affected eye
- In dark or light
- gating or hypersensitivity at LGN, deafferentation

Page N. et al JNNP1982;45:7-12

Occipital Epilepsy

- Blind during the attack
- Postictal headache
- Mis-diagnosed as Migraine, completely different from migraine
- Rx Carbamazepine

Panayatopoulos C.P. JNNP 1999;66:536-540

Visual Symptoms in Children with Reading Problems

- Words Jump
- Words Blurr or overlap
- Double vision
- Lines skipped (losing place)
- Letters missed

Seminars

Retinal Disease- when do I call the Neurologist?

David Taylor, London

A. Batten's Diseases
- CLN3, history
- Visual deterioration
- Mental deterioration
- Seizures
- Diagnosis
  - Vacuolated lymphocytes
  - Electron microscopy
- Gene structure
- Prenatal diagnosis
- Treatment?

B. Leber's Congenital Amaurosis
- Diagnostic criteria
  - Blindness from birth
  - Severely attenuated ERG
  - Absence of another retinal or systemic disorder
- Ocular findings
  - Nystagmus
  - Non-recordable ERG
  - High Hypermetropia
  - Eye rubbing
  - Variable retinal findings
  - Visual deterioration
- Associated systemic abnormalities
- Leber's Genes
- Differential diagnoses

C. Peroxisomal disorders
- Zellweger's syndrome
- Neonatal Adrenoleucodystrophy

Infantile Refsum's disease
PEX gene mutations

D. Alstrom's disease
Poor Vision from birth
Nystagmus
Cardiomyopathy
ERG_
Hypermetropia
Obesity
Deafness
Diabetes Mellitus

E. Joubert's syndrome
Neonatal tachypnoea
Developmental Delay
Cerebellar Signs
Retinal dystrophy
Colobomas

F. Jeunes Syndrome
Asphyxiating thoracic dystrophy
Post-axial Polydactyly
Retinal Dystrophy
Kidney & liver disease

G. Carbohydrate-deficient glycoprotein syndromes
Type1a, PMM2 mutations
Cerebellar & brainstem atrophy
Stroke-like episodes
Dysmorphia, fat distribution & skeletal
Diagnosis
CDG1b may be treatable

H. Many others, not discussed here!
Biedl Bardet, Hallervorden Spatz, Oxalosis

Plenary Lecture

Does My Baby See, Doctor?

The apparently blind child

David Taylor
London

Presentation
History
Vision
- Onset?
- Progression?
- Photophobia?
- Day Blindness?
- Light Staring?
- Night Blindness?
- Eccentric Viewing
- Eye Rubbing?
- NystagmusSocial Circumstances

Hearing
General Health and Development
Family History
Congenital anomalies

Simple clinical assessment -it's a game!!
Visual responses
Fields
Colour vision
Pupils
Refraction
IOP
Fundoscopy
Slit Lamp examination is possible at any age

Family History and examination of relatives
A look at the whole Child
VEPs and ERGs
- Suspected retinal disease
- All children with suspected poor vision
- Photophobia
- Watering without stickiness
- Systemic disease with known visual system disorders
- Congenital nerve deafness
- Nystagmus

- Suspected albinism
- Amblyopia Rx unimproved VA
- Bilateral poor VA
- Hysterical symptoms
- For visual prognosis
  i.e. Cerebral palsy
  Optic disc anomalies
- For visual assessment/sweep
  i.e. Cataract
- Bone disorders
  i.e. Osteopetrosis
- For genetic counselling

Technical aspects

Preparation....
Explain test and reas 3-4 minutes
Apply 5-6 electrodes 5 minutes
Recording....
i Pattern stimulation
Range of checksizes (25'-7deg) 15-20 minutes
ii Flash stimulation(ERG & VEP)
Remove electrodes 3-4 minutes
Information concerning test Variable

Total test time 30-35 minutes

MRI Scan

Normal ERG, abnormal VEP
Infant with ONH or coloboma
Compound nystagmus
Proptosis.
?Heminanopias
Cranial nerve palsy

CT Scan

Bone diseases, Proptosis.

"Subtle" causes of uniocular reduced VA

- Cataract
- Foveal hypoplasia
- Macular pigmentation
- Retinal disease
- Optic nerve defects -ONH
  -OA
  -pit

- Refractive causes
- Nystagmus

Normal fundi with bilaterally poor VA
-peripheral causes

- Chiasmal & ON compression
- Nystagmus
- CSNB
- Cone dystrophy
- Rod/ Cone dystrophy
- Inadequate examination
- Pseudoblindness
  - –DVM
  - –Saccade Palsy

-"Central" causes

- Acquired
  - –NAI
  - –Infections
- Perinatal
  - –PVL
  - –HIE
- Developmental
  - –Lissencephaly
  - –Pachy/polymicrogyria
  - –Porencephaly
  - –Holoprosencephaly
  - –Dysmyelination
  - –Metabolic disease

Brain Blindness-symptoms and signs

–Poor VA
–nystagmus
–Normal eyes
–Normal pupils
–Perceptual problems
–Blindsight?

The wider Issues

- The extended family
  - –Sibs
  - –Grandparents
- What does the future hold?
- There is always treatment!
- Optimism is better than Pessimism!
- Early intervention works!