Health in Adults with Cornelia de Lange Syndrome

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Overview

- Comprehensive Health Assessment - what is involved
- Common health problems
- Preventative measures
- Screening
- Adult CdLS clinic – brief look
Characteristics of health conditions in people with ID

• Many health conditions in people with intellectual disability are:
  – Multiple - multiple conditions seen at younger age than in general population
  – Complex
  – Chronic – and occur at younger age than in general population - e.g. dementia, osteoporosis
Preventing health problems

• Many of these problems are more common or become worse with ageing

• Communication difficulties can make it difficult for people with developmental disability to recognise and communicate pain, discomfort and other symptoms of ill health

• Identifying these problems therefore often requires screening (testing) of people with developmental disabilities

• Being aware of these conditions enables treatment or prevention of further disease
The Health Assessment.

- What are they?
- How often and what should this involve?

- History
- Examination
- Investigations
- Diagnosis
- Recommendations
Assessment of Adult with CdLS

- Full diagnostic workup
  - differential diagnosis of problems
  - assessment of co-morbidities
- This will identify areas to be addressed in a management plan
- Evaluation of current and future treatments
Comprehensive medical assessment

• There could be any number of morbidities but a systematic approach and mindfulness of common causes particularly in those with ID/CdLS will mean less likely to miss a physical cause.
• General practitioner
• General physician (endangered species)
• Geriatricians/rehabilitation physicians
• Clinics specialising in medicine of people with intellectual disabilities.
  ▪ Developmental Paediatricians
  ▪ Developmental disability physician

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Conference 2011
Clinical History

- Informants
  - Patient (acquiescence and suggestibility)
  - Family members (background, bias, selectivity)
  - Support workers/advocates (as above)

- Old notes and documents

- Clinicians: Specialists and allied health workers
Make the most of your Medical visits:

- Book a long appointment
- Attend with knowledgeable support person
- Find out if the surgery and the examination table are easy for you to access
- Before the visit, write down all the questions you want to ask and issues you would like to discuss
- Take all your current medications and a record of your medical history
- Bring patience, picnic, change of clothes
Medical examination

• Medical examination
Medical examination

• Physical examination: specific or general
• Full medical examination = clothes off.
• Mental /Psychological assessment
Investigations

- Blood tests: FBC, Fe studies, biochemistry, Lipids glucose, Hepatitis serology, TFT, B12, folate, Vit D H Pylori serology or breath test. Karyotype/CGH Array and metabolic screen
- Urine for microscopy and culture
- ECG/echocardiography
- Imaging: u/s, Xray, CT, MRI
- Bone densitometry
- Screening tools developed for people with ID
Management

• Specific problems
• Screening – following standard protocols
  • Women’s health
  • Men’s health
• Preventative measures
  • Immunisations
  • Lifestyle modification
  • Environmental changes
  • Behavioural management
Conditions to be aware of in older people

- Coronary heart disease
- Stroke
- Cancer
- Hypertension
- Diabetes
- Vision / hearing problems
- Arthritis
- Osteoporosis/vitamin D def.
- Falls
- Dental problems/tooth loss

- Poor nutrition
- Impaired immunity
- Urinary incontinence
- Constipation
- Hypothyroidism
- Skin fragility
- Depression
- Dementia
- Polypharmacy / iatrogenic illness
Behavioural concerns

• Underlying physical problems (eg pain, discomfort, thyroid disease)

• Environmental factors (eg change of carer, loss of family or friends due death, illness or moving away)

• Management:
  – Behavioural +/- medications
  – Referral to a psychologist
  – Referral to a psychiatrist
Mental Health Issues in PWID

- Self injurious behaviours
- Autism/autism spectrum disorder
- Depression

- Challenging behaviours
  - Psychiatric illness
  - Physical illness
  - Psychosocial problems
  - All of the above.
Medical Conditions common to PWID

- Epilepsy
- Respiratory problems
- Oral health – poor dentition
- Malnutrition
- Musculoskeletal - osteoporosis
- GIT
  - GORD +/- H.pylori infection
  - Constipation
  - Surgical
Sensory impairment

- Vision impairment 7 times higher than general population (Warburg 1994)
  - Cataracts are common and can present with behavioural changes
  - Includes problems with near vision or far vision
  - Degenerative changes (ie worsen with time)

- Deafness 11 times higher than general population
Vision impairment

- Routine assessment by an eye specialist (ophthalmologist) or optometrist every 5 years from age 45 years is recommended.
- Obviously if there is an existing vision problem more frequent reviews may be necessary.

Hearing

- Formal hearing test every 5 years from age 45 years (or earlier if there appears to be problems)
- The Australian Government Hearing Services Program:
  - Phone: 1800 500 726
  - Office of Hearing Services Website: www.health.gov.au/hear
Iatrogenic

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<td>Antidepressants</td>
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<td>Cannabis</td>
<td>Lithium</td>
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Polypharmacy
Prevention – medication problems

• Review by GP of all medications every 3-6 months

• Webster packs and dosette boxes are important particularly when there are multiple carers and support staff
Life expectancy in people with intellectual disability

- Life span increasing but still less than general population
  - up to 20 years less (Bittles et al. 2002)

- Mild intellectual disability & few medical problems – near average life expectancy
CdLS Clinic

• 2-4 clinic days a year
• Comprehensive medical assessment
  – Specific complaints
  – Screening – following standard protocols
  – Prevention
  – Assist in coordinating patients care locally
• Initial appointment 2 hours with a 3 month follow-up
• Annual medical reviews offered at DDHU
CdLS Clinic

• 15 patients seen
  - 14 CdLS diagnosed prior to initial visit
  - One patient did not have CdLS – CHARGE syndrome
  - One new diagnosis made
  - 2 patient with features suggestive of CdLS at general clinic

• Outreach consultation days –
  - Melbourne = 7 consultations
  - Adelaide = 10 consultations
CdLS Clinic at Health Unit

- 9 female 6 male with CdLS

- Age range 13 years – 47 years  Average: 28.7yrs

- 4 patients - Genetic diagnosis confirmed
  11 patients – no genetic diagnosis
CdLS Clinic
Level of Intellectual Disability

Mild, 1
Moderate, 2
Severe, 9
Profound, 3

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Clinic patients cont.

• Main clinical problems
  - Behavioural concerns
  - GORD
  - Constipation
  - Vision
Weight of Patients with CdLS

• Weight
  – Underweight = 3
  – Healthy weight range = 10
  – Overweight = 2

• The 2 patients PEG fed in healthy range
Clinic patients - Challenging behaviours

- Sleep disturbances – 5 patients
- Self-injurious behaviours – 12 patients
  - head banging
  - wrapping
  - skin picking/scraping
- Ritualistic/ocd = 9 patients
- Aggression = 6 patients
- Stripping = 3 patients
General impressions at clinic

• GIT symptoms are ongoing
  • GORD
  • CONSTIPATION
  • Nutrition well attended

Women normal menstruation

• Vitamin D levels from 7 patients from initial blood tests
  – Normal = 4 patients
  – Mild insufficiency between 40-50ng/mil = 3 pts
Summary

- A requirement for the diagnosis or exclusion of a medical or psychiatric condition requires a comprehensive medical assessment and the use of appropriate assessment tools specific to the population of people with intellectual disability.

- Gastrointestinal
- Neurological
- Epilepsy / trauma
- Osteoporosis and 
- Sensory impairment
- Endocrine disorders
- Toxins
- Nutritional disorders
- Infection
- Cardiovascular disease
Health Guidelines

• Developed by the Physical Health Special Interest Research Group of the International Association for the Scientific Study of Intellectual Disability (IASSID)

• Ratified by WHO in 2006
Acknowledgements

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