Roles of Community Paediatrician in Hearing Loss. Assessment of Emerging Developmental concerns

Introduction

- Heterogeneous nature of Hearing Loss.
- Scope of this talk - role of community paediatrics and other agencies in assessment of development and the aetiology of hearing loss. Rationale for paediatric assessment. Overview of common tests. Importance of confidentiality.
- Overview of common presentations for hearing loss associated with developmental concerns, where to get more information and what to do next.
- Importance of differentiating isolated (non-syndromic – vs syndromic hearing loss with case histories
- How to identify issues which suggest futher investigation would be helpful. Medical red flags.

Developmental difficulties in hearing loss

Known cause of hearing loss which may be associated with developmental difficulties. Eg congenital CMV, genetic.

Further testing

Developmental Assessment reveals Additional concerns

Concern raised by parent or other professional TOHI etc

Environmental factors – Additional issues, CP/ sleep/ Resources/ Equipment

Role of Paediatrican

- See Newly Diagnosed, progressive loss, new developmental concern (<6 or ref via suggestion ed psyche – if medical concern), new possibly related medical issue eg renal problem, seizures, visual problem in child who does not have a confirmed cause of non-syndromic hearing loss.
- Children are seen at New Dingley https://youtu.be/TBk38VPwRWs

Role of paediatrician

History including birth and prenatal, family history

Examination – looking for distinctive facial features, ear malformations, neurocutaneous changes ears and neck pits tags sinuses.

Developmental assessment under 5s tools such as Schedule of Growing Skills II, drawing, social communication

Investigation – depends on child see below.

Role of Paediatrician - Investigations

Assess for underlying cause – and investigate, detailed history including history of hearing loss and family history.

In under 5s direct developmental assessment.

Every child – family audiograms, CMV and eye specialist.

Every child diagnosed at birth or other concerns - ECG

Moderate and worse bilateral – Genetics - Connexin 26 testing (AR pattern no developmental concerns)

MRI

Permanent conductive CT,

Additional developmental concerns or other congenital anomalies – consider array CGH.

BP and urinalsis (all diagnosed after 6 years, other malformations or family history.

History

When hearing loss identified any other concerns

Mothers health in pregnancy

Full family tree asking about hearing,vision, thyroid/ kidneys, learning, other rare disorders.

Different coloured eyes

History of early development – walking, dizzyness, tinnitus.

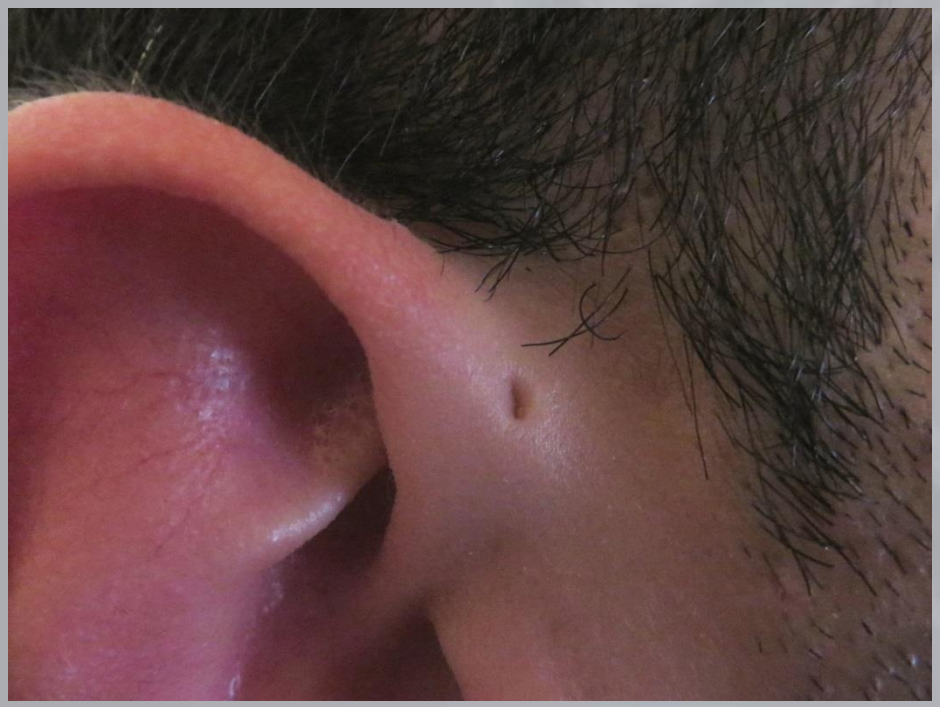
Sleep and activities of daily living

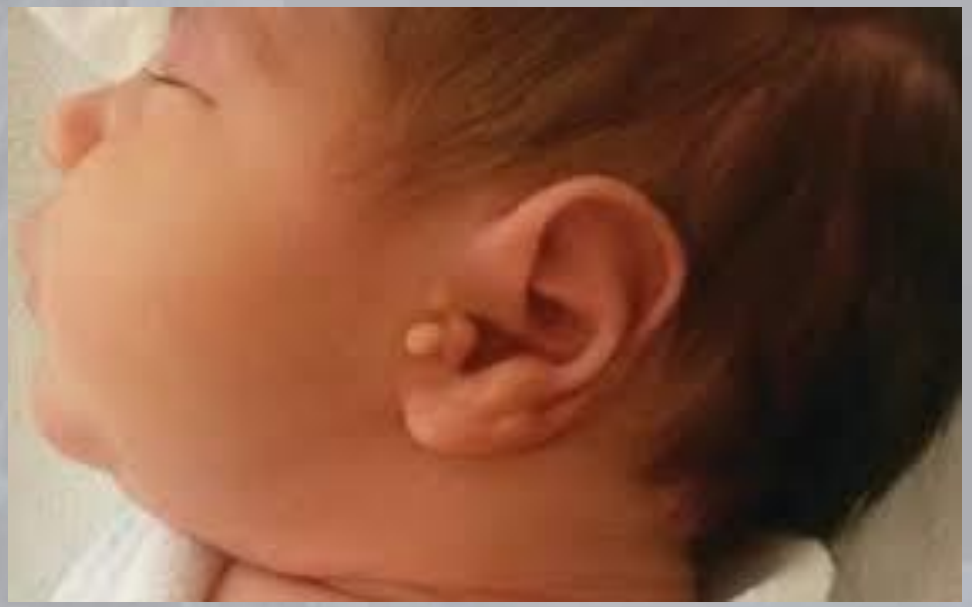
Examination

Head to toe.

Including growth and head circumference

Ear pits tags and sinuses.

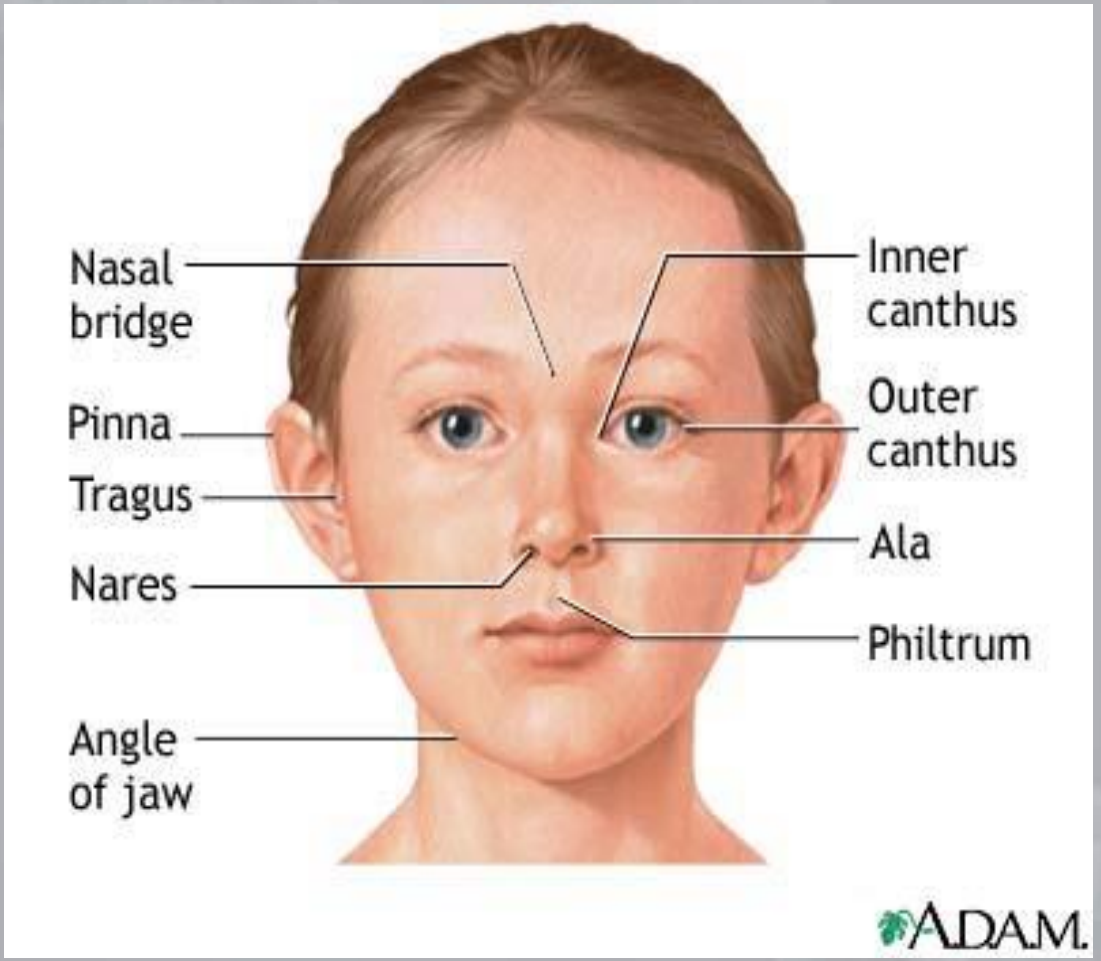




Facial Appearance – What to look for?

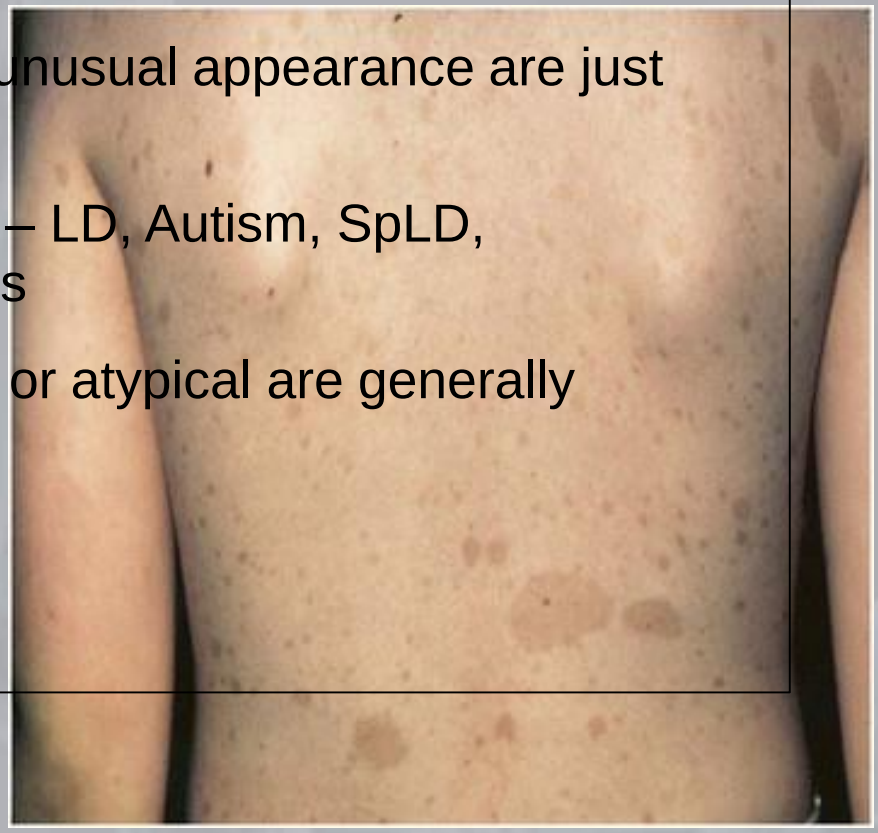
- Face develops in first trimester. Closely linked to brain development.
- Eyes – distance, slant of palpebral fissues, epicanthric folds.
- Ear size position and rotation
- Mouth, size,
- Nasal philtrum – should have a groove
- Thickness of upper lip
- White forelock,

Facial Appearance – What to look for?

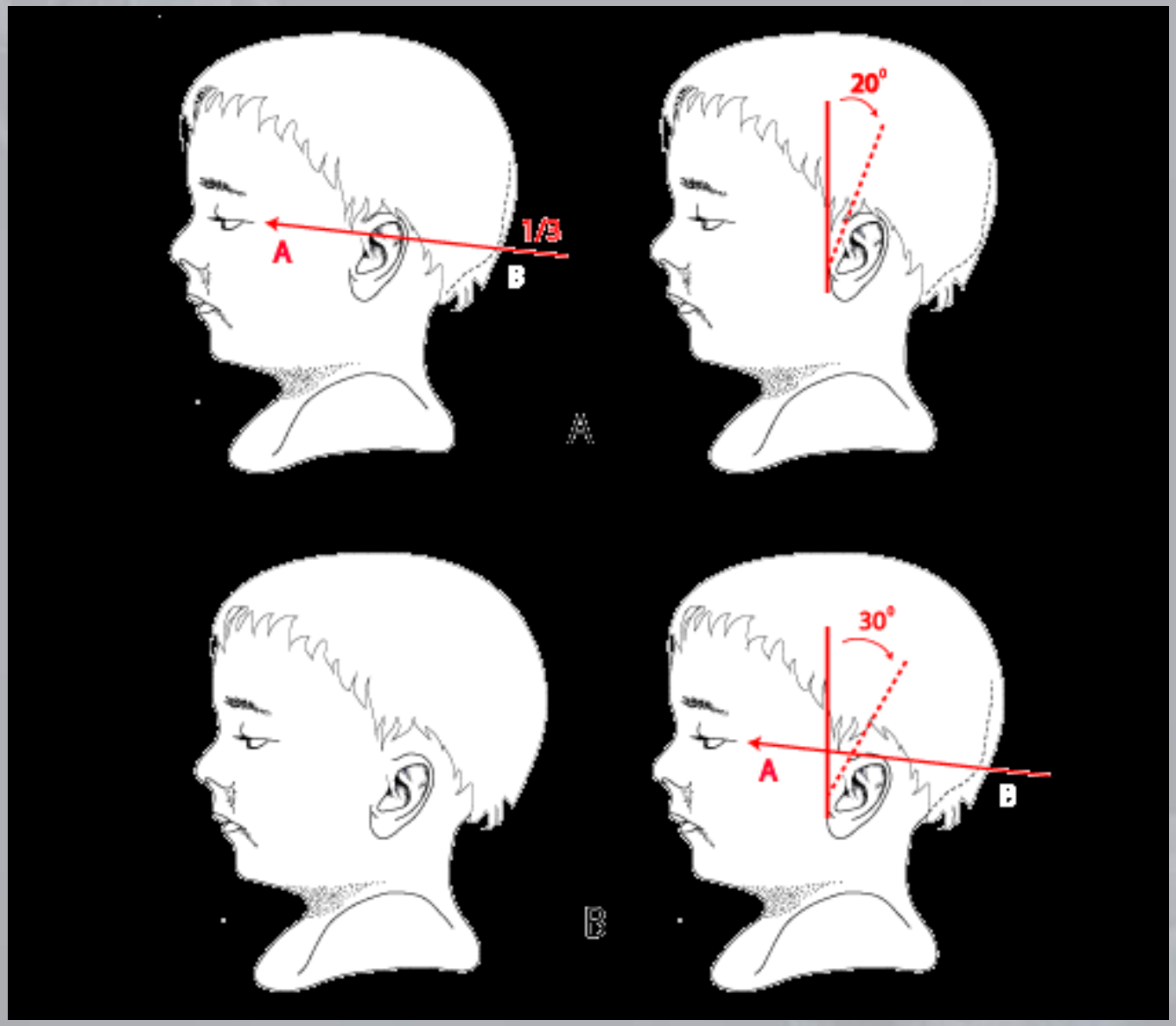


Physical Appearance – signs to look for

- Varies from condition to condition
- long face, prominent ears
- Small head – microcephally, large head macrocephaly,
- Neurocutaneous markers – multiple café au lait
- Comparison to family – some children with unusual appearance are just following their families pattern but..
- If similar looking family members also have – LD, Autism, SpLD, neuropsychiatric problems consider genetics
- Take care with language – striking, unusual or atypical are generally acceptable.



Facial Appearance – what is typical



What is Development?

What is Development?

•Dynamic, ongoing process that continues throughout life.

Reacting to sensation → Making sense of

/ being dependent information/ responding in a planned,

organised and independent

Fashion

•Orderly, sequential, and predictable,

•Builds on earlier learning/ skills

–From simple to complex

Factors Affecting Development

•Biological influences:

–Inherited characteristics such as cognitive potential

–Antenatal and perinatal factors

–General health

–Vision and hearing

•Environmental influences

–Opportunities

•Sensitive and supportive parent/family

•Educational placement

–Disadvantages

•Social and economic deprivation

Developmental Milestones

- Set of functional skills or age-specific tasks that most children can do at a certain age range.
- Convenient guidelines to assess the rate and the pattern of developmental progress.

*Many aspects of development are continuous and stages may overlap.

Developmental Domains

- Gross motor
- Visual perception and fine motor
- Speech, language and communication
- Social behaviour and play
- *Hearing and vision*

Gross Motor Skills

Fine Motor Skills

Communication skills

Social/ Emotional Skills

Case History

Sensorineural hearing loss diagnosed at birth, urine CMV positive.

History of mum being unwell in pregnancy – fevers joint pain.

Guthrie card +ve.

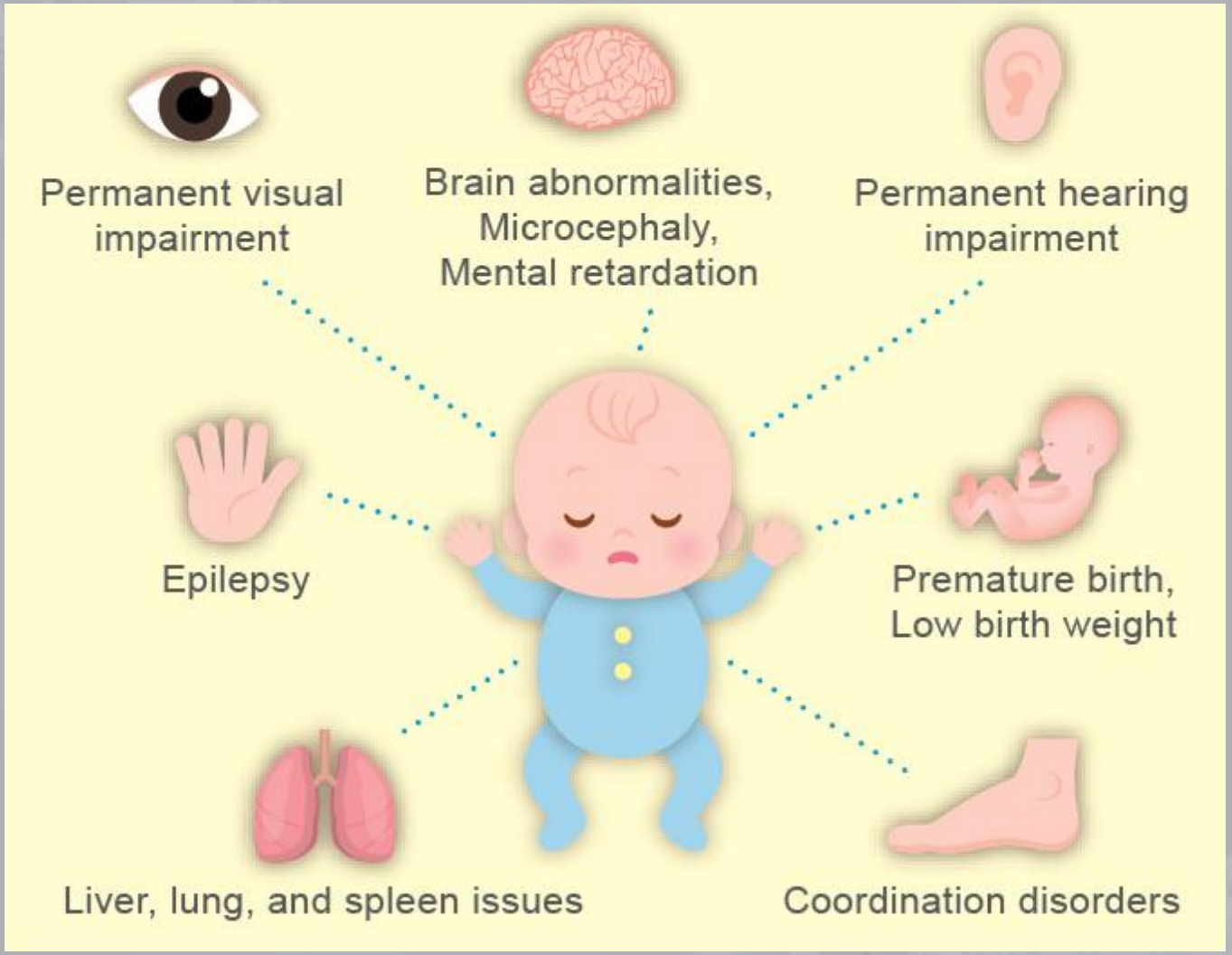
Examination: small at birth small head circumference.

Diagnosis confirmed as CMV is known to be associated with other issues.

Opthalmology (eye specialist review) and regular developmental surveillance. Normal neurology exam. Walking slightly delayed but normal

Developmental assessment signs of Global developpmental delay, concerns about autism.

Congenital CMV



Case History

20 month old child with congenital sensorineural hearing loss and mild global delay on paediatric assessment.

As Hearing loss was associated with mild global delay an array CGH was organised in addition to connexin 26 mutations.

The test revealed a rare disorder. Deletion Chromosome 10 p including within exon 6 of the GATA 3 gene – HDR (Barakat) syndrome
Renal problems hearing loss and hypoparathyroidism
Denovo mutation. Many different associations,
Renal US fine and bloods – having surveillance for hypoparathyroidism, will have repeat scans.

Advantages of identifying underlying diagnosis.

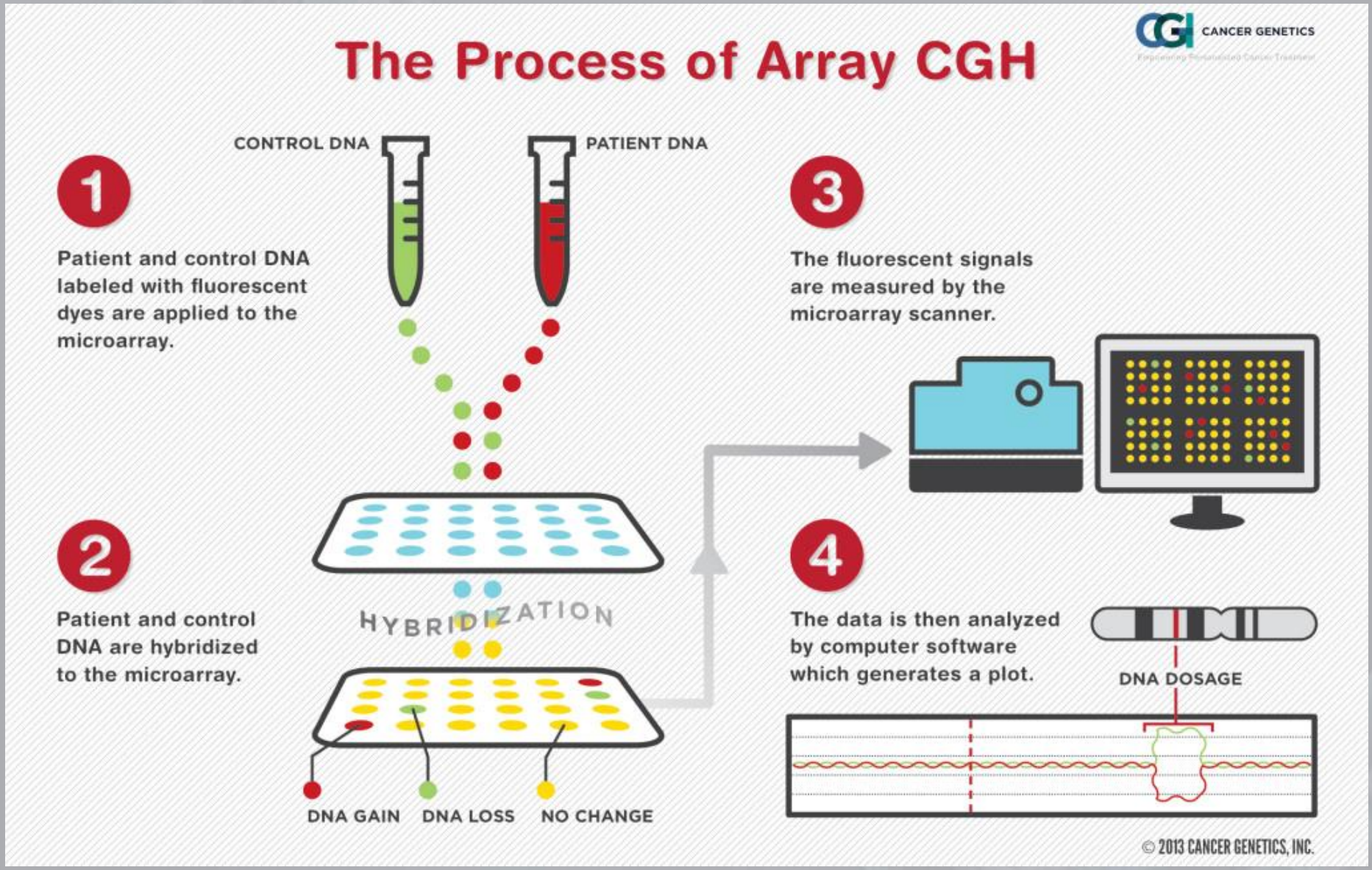
Able to identify and manage other associated medical problems.

Eg in HDR syndrome

Showing 31 of 31 | View Less

Medical Terms	Other Names	Learn More: HPO ID
100% of people have these symptoms		
Hypoparathyroidism	Decreased parathyroid hormone secretion	0000829
Progressive sensorineural hearing impairment		0000408
Renal dysplasia		0000110
30%-79% of people have these symptoms		
Hydronephrosis		0000126
Hypocalcemic seizures	Low calcium seizures	0002199
Parathyroid hypoplasia	Small parathyroid glands [more]	0000860
Polycystic kidney dysplasia		0000113
Renal insufficiency	Renal failure [more]	0000083
Unilateral renal agenesis	Absent kidney on one side [more]	0000122
Vesicoureteral reflux		0000076
5%-29% of people have these symptoms		
Diabetes mellitus		0000819
Septate vagina	Double vagina	0001153
Uterus didelphys		0003762
1%-4% of people have these symptoms		
Abnormal heart morphology	Abnormality of the heart [more]	0001627
Abnormality of T cell physiology		0011840
Aplasia of the uterus	Absent uterus [more]	0000151
Cleft palate	Cleft roof of mouth	0000175
Psoriasiform dermatitis		0003765
Rod-cone dystrophy		0000510
Sensorineural hearing impairment		0000407

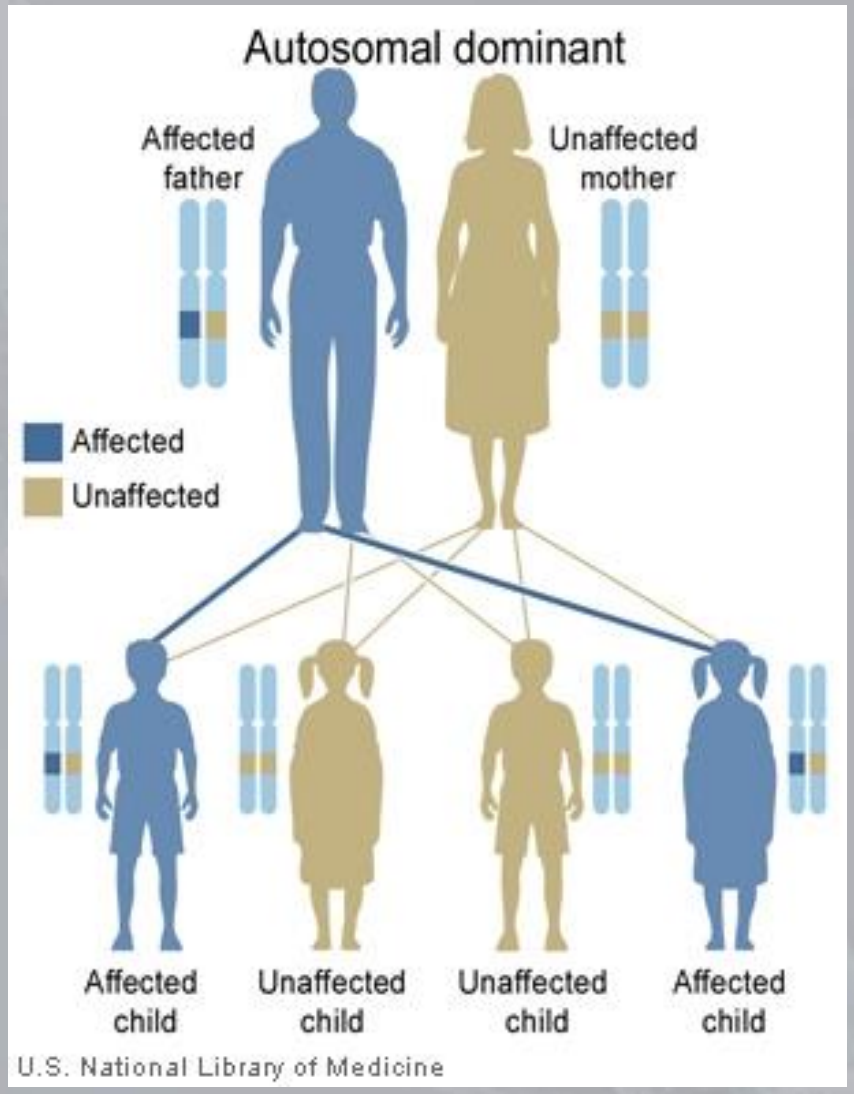
Micro deletions and duplications – Array CGH



Array CGH

- Array CGH – usually gives either normal or potentially abnormal result
- Indicated for Deafness plus developmental delay or congenital abnormalities.
- Relatively new technique, still working out what is normal, therefore may get result of uncertain significance.
- Reported as duplication (extra material), deletion (missing material) compared with international anonymised data base, see if others with same changes
- Investigated by taking samples from parents, probe for change to see if carried.
- More likely to find dominantly inherited conditions, but not great for Dominant non-syndromic hearing loss. Can refer to geneticist for more complex testing if wished.

Dominant Inheritance.



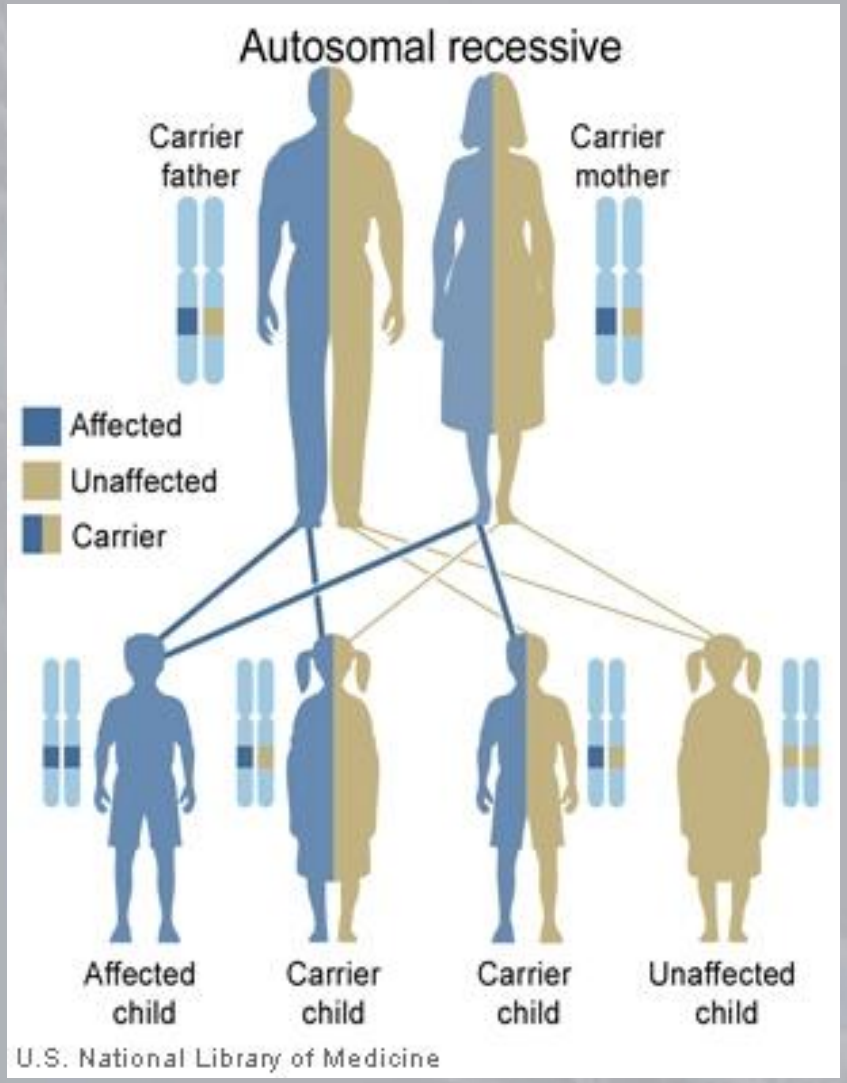
Case History - ? Additional problem

- **Identified at birth severe hearing loss no family history**
- **Saw paediatrician – CMV negative, connexins ordered.**
- **Connexin mutations identified autosomal recessive Connexin 26 mutations. – not normally associated with developmental concerns. But family history in cousin of ASD**
- **Progressed to Profound had cochlear implant at 16 months**
- **Seen at 24 months concerns about speech delay concerns from multiple professionals SLT and implant team re progress.**
- **Increased developmental surveillance – appeared to be ? Concerns about social communication - ? Additional disorder recommendations made**
- **On review significantly improved speech and social communication**

After time and implementation of strategies – now felt to be on a normal trajectory

But will be kept under review.

Autosomal Recessive inheritance



Case History

3 year old boy – profound progressive SN hearing loss diagnosed as CMV early in infancy.

Cochlear implant – seen for developmental review.

Concerns about sleep behaviour and development

? Additional social communication disorder,

Behaviour and social communication worse with sleep, seen for review mixed presentation mum and child exhausted.

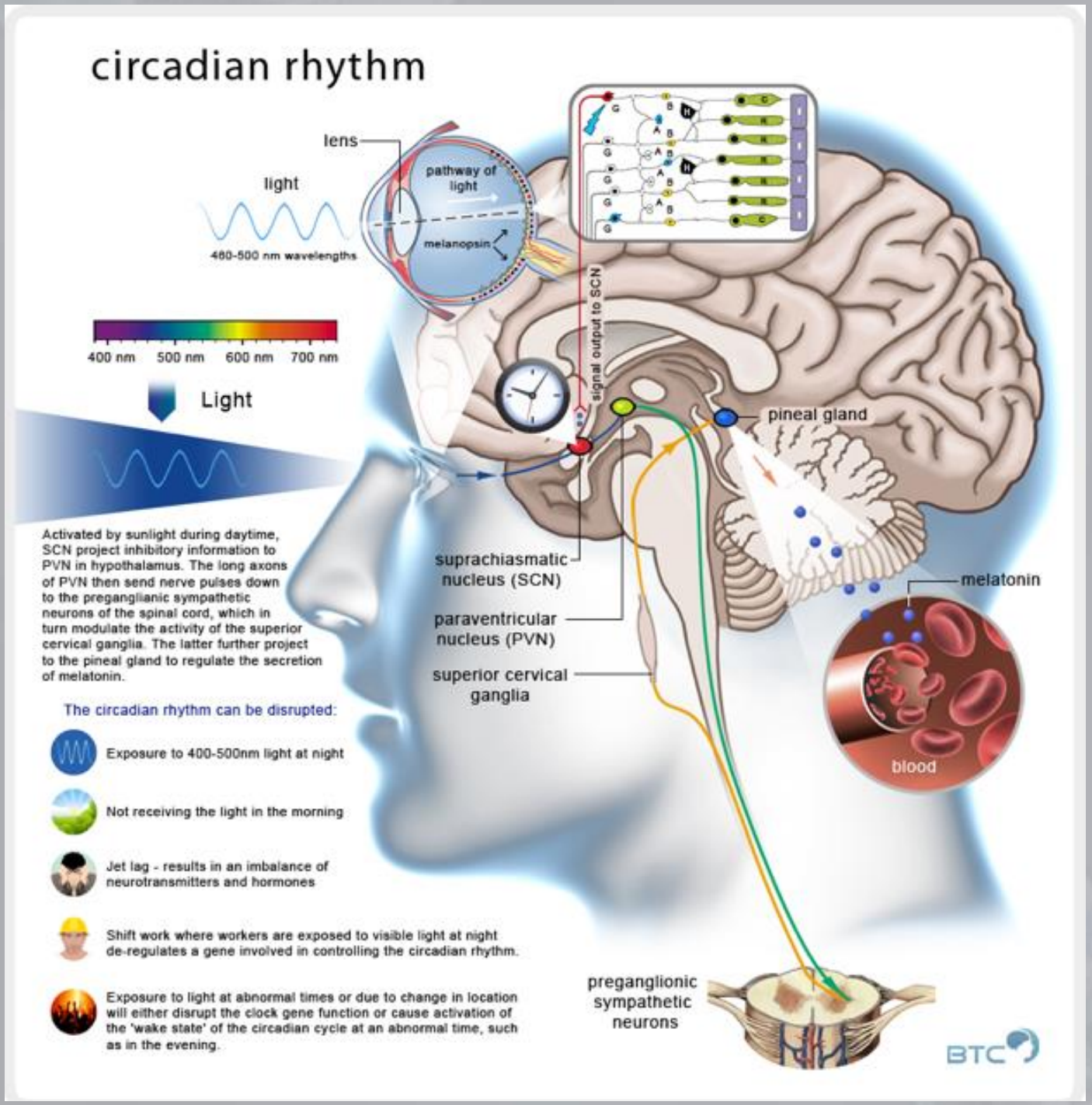
Seen for review of sleep and development.

Discussed sleep hygiene and environment. Using Ipad and Blue light. Unsettled after processors removed scared of the dark. Mum staying with him, waking every hour on the hour.

? Linked to sensory deprivation discussed sources of help and support

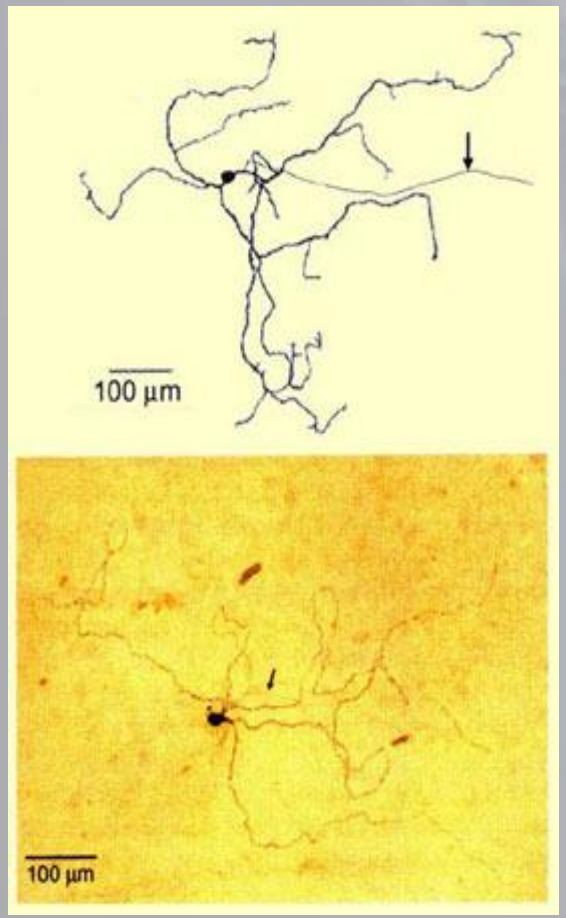
Parenting special children sleep course

Thought through basics of sleep and sensory based approach


circadian rhythm
lens
light
pathway of light
melanopsin
460-500 nm wavelengths
400 nm
500 nm
600 nm
700 nm
Light
signal output to SCN
pineal gland
melatonin
blood
suprachiasmatic nucleus (SCN)
paraventricular nucleus (PVN)
superior cervical ganglia
preganglionic sympathetic neurons
Activated by sunlight during daytime, SCN project inhibitory information to PVN in hypothalamus. The long axons of PVN then send nerve pulses down to the preganglianic sympathetic neurons of the spinal cord, which in turn modulate the activity of the superior cervical ganglia. The latter further project to the pineal gland to regulate the secretion of melatonin.
The circadian rhythm can be disrupted:
Exposure to 400-500nm light at night
Not receiving the light in the morning
Jet lag - results in an imbalance of neurotransmitters and hormones
Shift work where workers are exposed to visible light at night de-regulates a gene involved in controlling the circadian rhythm.
Exposure to light at abnormal times or due to change in location will either disrupt the clock gene function or cause activation of the 'wake state' of the circadian cycle at an abnormal time, such as in the evening.
BTC

Role of intrinsically light sensitive retinal ganglion cells

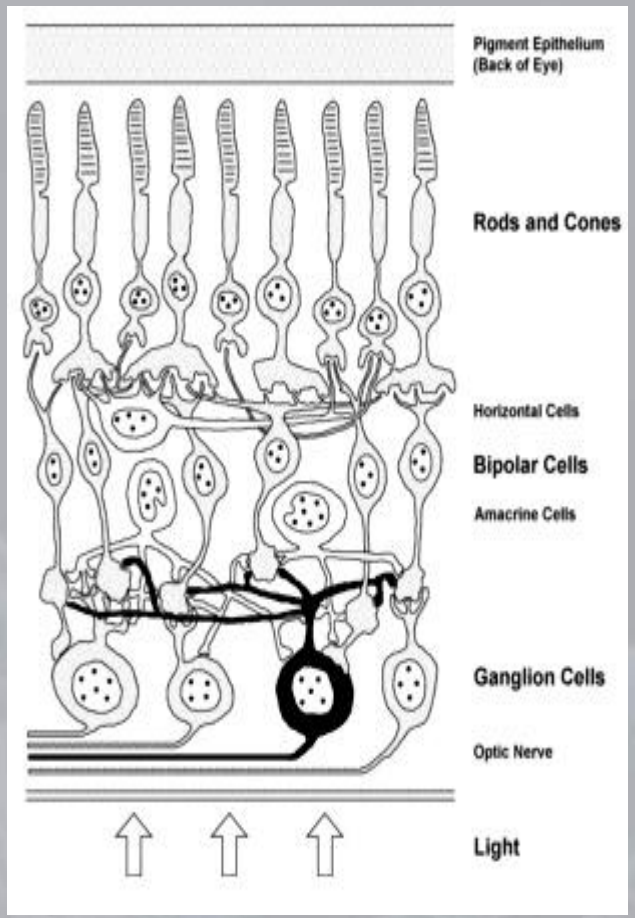
In 1991 – Russell Foster discovered Intriniscally photosensitive retinal ganglion cells



Express melanopsin

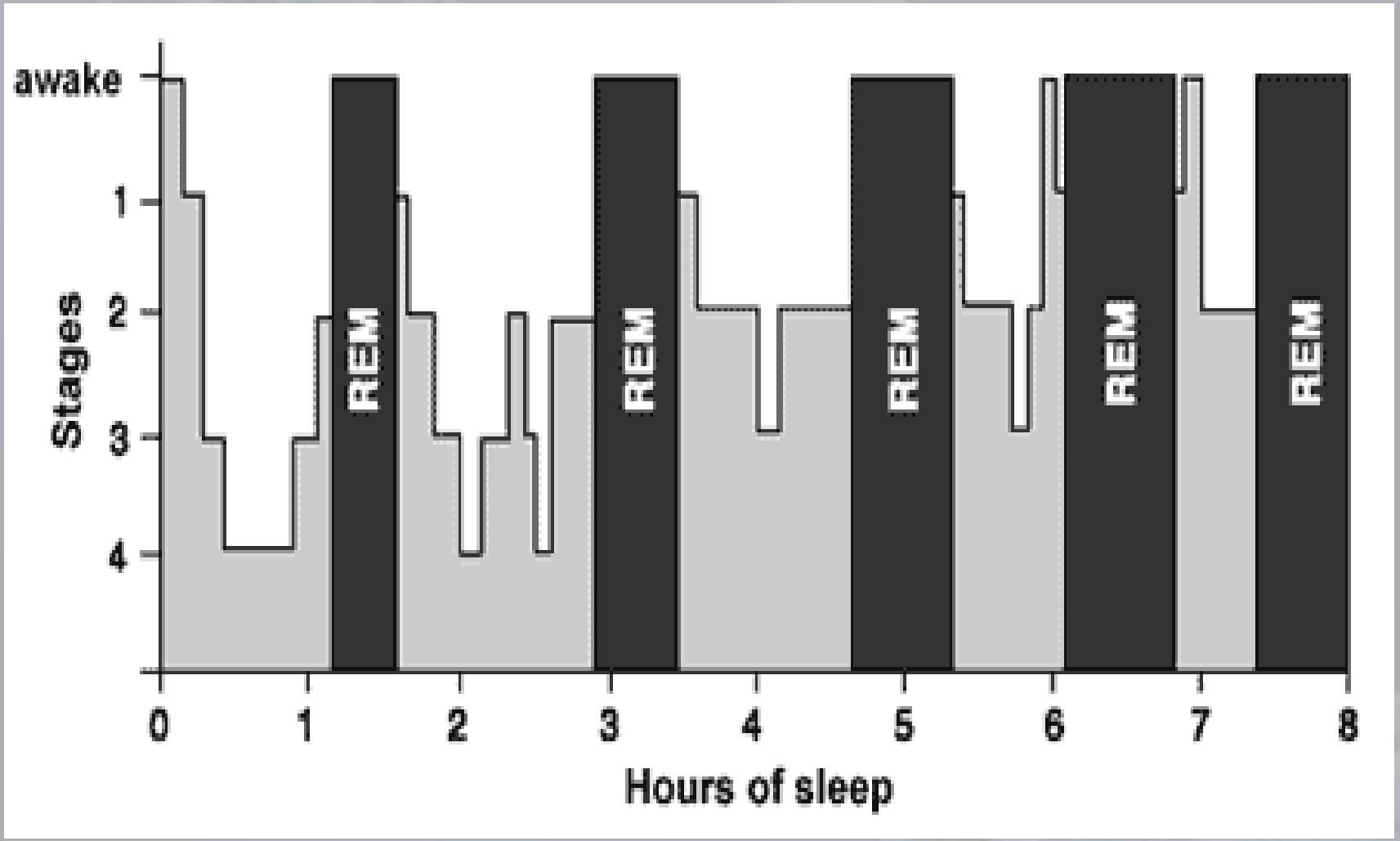
Signal inhibiting melatonin secretion

Peak sensitivity 460 and 484 nm (Blue).



Sleep Architecture

Sleep Architecture – Hypnogram (EEG,EOG,EMG)



Environmental change

Working in Partnership

Mum attended course,

Did some research and

Found Sleep tight night

Time teddy.

Discussed option of

Meds if not sucessful,

Not needed,

Outcome

Reviewed in 4 months

Sleeping better with environmental change,

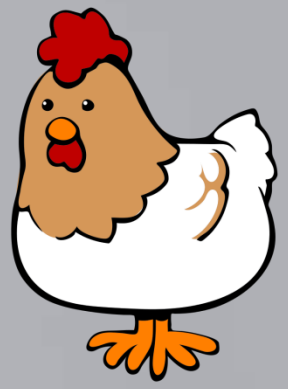
Decreased concern about behaviour and social communication.

Doing better developmentally but still some concerns

Plan keep development and social communication under review.

I learnt another strategy to help other clients.

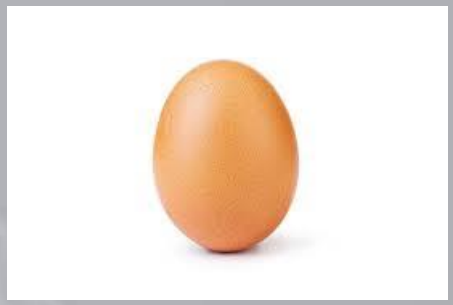
Hearing loss and social communication – chicken or egg?



Social communication difficulties can arise from hearing loss –
However some deaf children have social communication disorders

How do we determine which it is?

When do problems occur?
Pervasive or situational
What are the non- verbal skills like?
Eye contact gesture compensation for difficulties with hearing loss and speech.
Imagination
Interaction with adults vs children
Access to and use of aids?
Additional features not suggestive of hearing loss –
spinning flapping toe walking (can be normal until 2)
Drawing people at age appropriate stage?



Strategy for investigation – mild – moderate no complex features, younger child – local with paediatrician for hearing loss as part of team.
Moderate to severe or additional complexity – for tertiary assessment either deaf CAMHS or tertiary centre

Case global delay

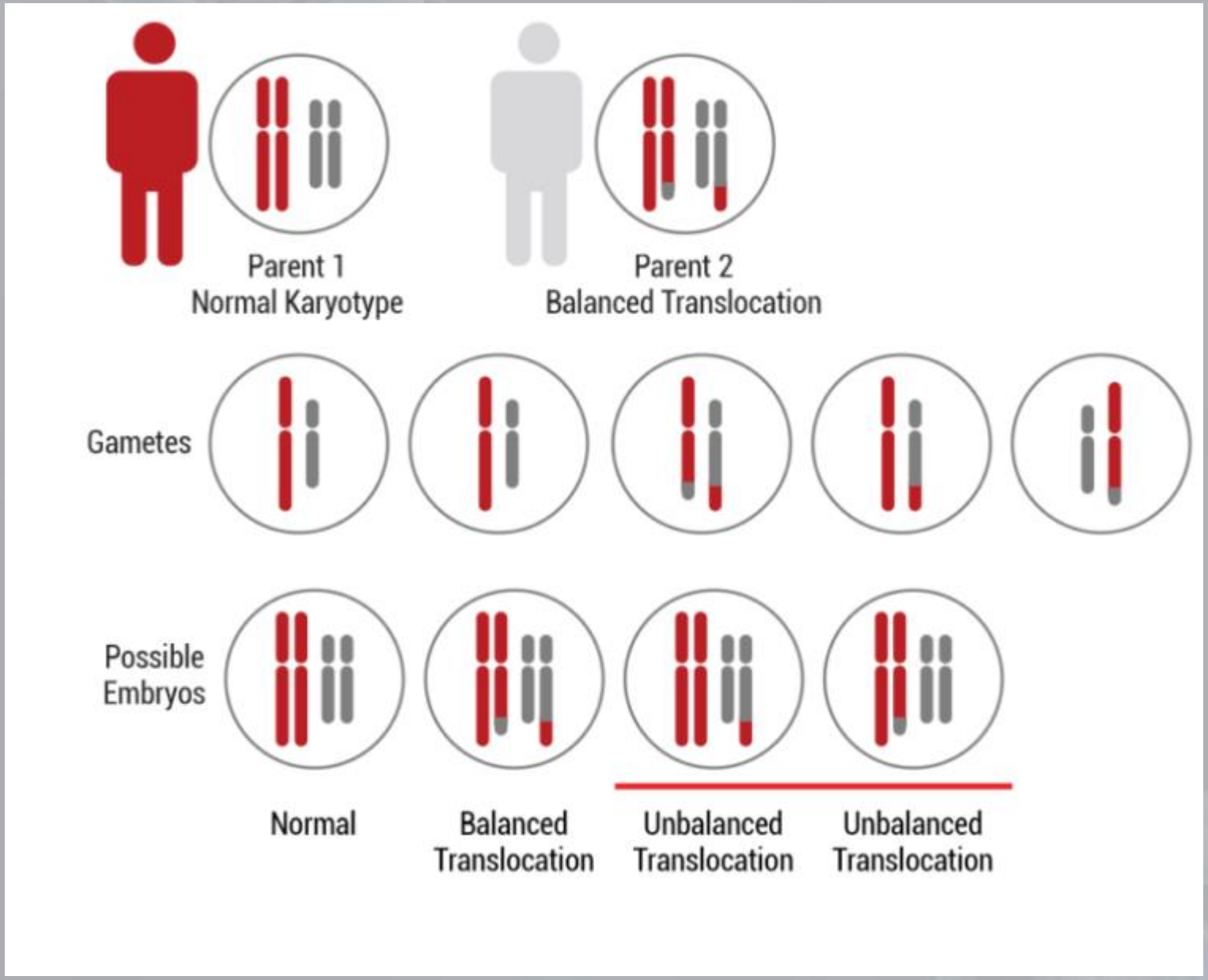
Presented with delayed global development and marked speech delay at 2 and a half the assessing paediatrician thought that her facial features looked unusual – thin narrow face small head.

Hearing test revealed permanent conductive hearing loss – managed by BAHA – bone implants now.

Chromosome ordered – unbalenced translocations extra piece of chromosome 18 and missing piece of another part of the chromosome. Known to be associated with developmental delay, hearing loss, growth hormone defiency and renal disease.

Management – developmental surveillance liase with education – for EHCP and special school, refer to endocrine team for growth hormone. Supportive footwear as foot malformation. Manage sleep difficulties. Renal ultrasound – cysts and small kidneys – recently presented with tiredness – abnormal renal function referred to Renal team

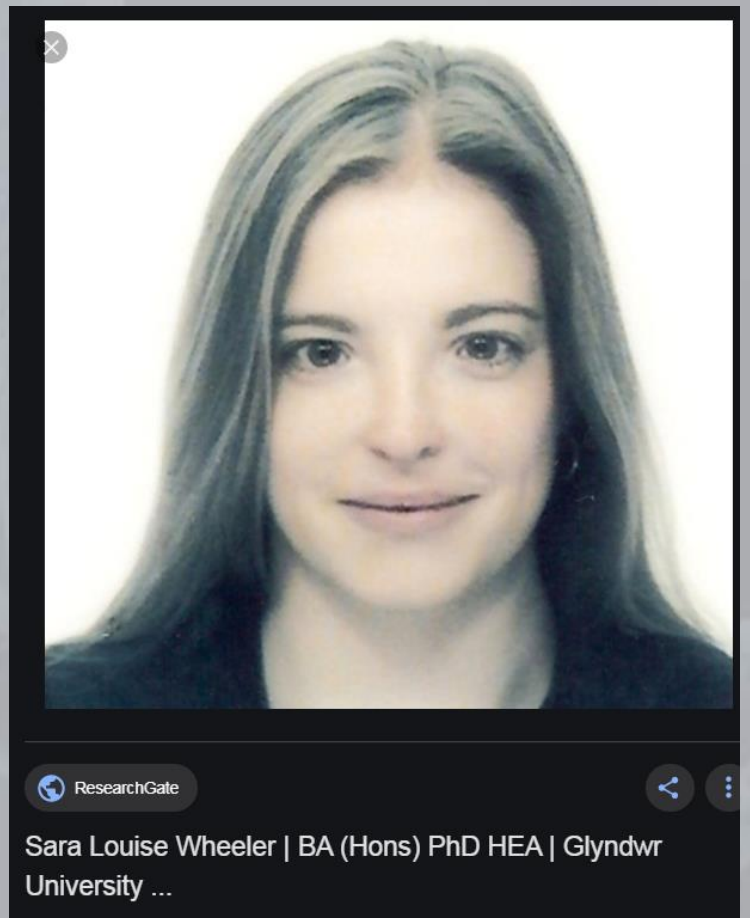
Unbalenced translocation



Pink Hearing aids and Purple Shampoo

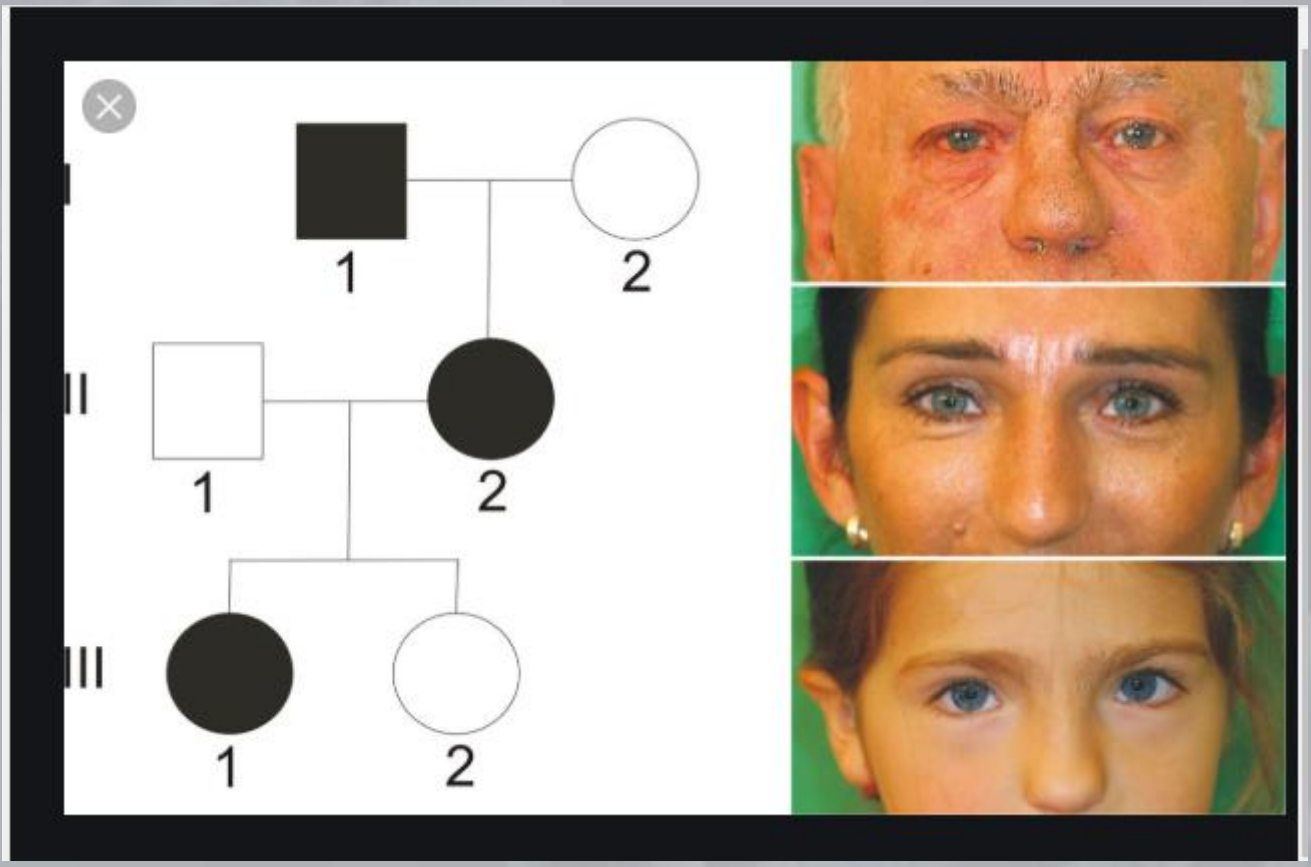
New referral in teens – low frequency reverse slope SN hearing loss. Mother has congenital hearing loss, different coloured eyes and premature greying.

https://www.semanticscholar.org/paper/Pink-hearing-aids-and-purple-shampoo%3A-biographical-Wheeler/64500036ed087e9d205c097ad709d84ed6ea5a4a



Wardenburg syndrome

Genetic change – number of different types and genetic changes. SN hearing loss, eye colour, premature greying white forelock. dystopia canthorum



Summary

Joined up multidiscplinary working, everyones view is important.

Ideal is identification of cause of deafness where possible.

Investigate developmental concerns even where known non-syndromic cause.

Rule out other progressive hearing loss or visual problem. Rule out sleep or other medical problem.

By working together we can maximise childrens learning potential and life opportunities.