ABSTRACT

Genetic Counselling in Hereditary Deafness (Alan Fryer 1995)

Introduction:-

Information giving

The main questions that people ask when they come to a genetic counselling clinic have been summarised as follows :- Why did the problem happen? Will it happen again in future children? Will it be as bad or worse, if the problem does recur? Are there any tests?

Arnos et al. set up a genetic counselling clinic at Gallaudet University, Washington D.C. (a "liberal arts" university for deaf students). She states that *"the experience of the Genetics centre has shown that deaf individuals have a deep curiosity about the cause of their own deafness and the implications for future generations"* and *"the majority of the clients seen at the Gallaudet clinic seek genetic counselling for information about themselves and future children. The information does not usually affect the reproductive decisions that are made."*

Some deaf people view their deafness not as a disorder but as a cultural difference with their own language, practices, etc. Arnos believes that one needs to be sensitive in the use of terminology - e.g.. *'risk', 'abnormal', 'affected'* may be offensive to the deaf person who does not view their deafness as a disorder. *"Deaf couples may consider it a risk to have a hearing child"*! Words such as *'chance'* instead of *'risk'* and *'hearing'* and *'deaf'* instead of *'unaffected'* and *'affected'* may be appropriate.

Arnos - *"at present, genetic counselling in most centres in the USA is very underutilised by parents of the deaf and the deaf community as well."* In a survey, Arnos et al. discovered that *"most of the parents were not aware that genetic counselling was a way in which they could get more information about the cause of their child's deafness"* and *"many professionals and deaf people themselves may assume that genetic counselling will hold no benefits for them since many times it would not change their reproductive decisions". "Many benefits of genetic counselling are unrelated to reproductive decisions. Genetic evaluation can reassure, rule out the presence of more complicated syndromes and perhaps more importantly help the family to accept and adjust to the birth of the deaf child".*

Arnos has noted specific problems :-

(1) *"it has been our experience that deaf children often know very little about the medical history of their close family, especially if they had hearing parents"*; (they have missed out on hearing things stated by their parents in incidental speech).

(2) pedigrees may be complex

(3) deaf couple may have limited biology knowledge

(4) communication - takes time.

Workload patterns

Most referrals are for undifferentiated deafness - often no family history. Fraser states, *"despite the multiplicity of syndromes described, the great majority of cases due to single gene Mendelian inheritance are clinically undifferentiated"* and *"such hereditary cases are often isolated in the family".*

Diagnosis :- Accurate genetic counselling depends on a diagnosis. It is important to examine the patient for syndromic associations, perform relevant tests and analyse the pedigree, which can be complex. In assessment one needs to take consanguinity and paternal age into consideration.

Non-syndromal deafness (Isolated or Undifferentiated deafness). Most cases of hereditary profound hearing loss are sensorineural. Familial undifferentiated conductive hearing loss is likely to be autosomal dominant or X-linked. The most common type is otosclerosis.

What % of hereditary cases are syndromal? i.e. not associated with any other abnormalities? 15 - 30%

(a) **Childhood onset** - usual referral reason.

(b) **Late-onset** - *..."A major proportion of late-onset hearing loss is genetic"* (Gorlin) But what proportion? No reliable data available.

INHERITANCE PATTERNS - Autosomal dominant (AD), Autosomal recessive (AR), X-linked (XL), mitochondrial.
-relative proportions estimated to be each type

Types :- Morton 1991

AR 77% (59 - 85% in various series)
AD 22% (15 - 30% .....)
XL 1% (up to 5% - Reardon 4%)

(a) General problems in counselling each type :-

AD :- Variability, non-penetrance, new mutations, germinal mosaicism, paternal age effect.
AR :- Consanguinity, heterozygote detection, heterozygote frequencies in different populations, HETEROGENEITY.
XLR :- Carrier detection, new mutations etc.

As far as the AR types are concerned - How many AR genes are there? Different studies make different estimates:- in essence nobody knows!

Morton (Hereditary Deafness Newsletter 1991) states that *"the search for non-syndromal loci will be difficult if the number of loci is as large as genetic studies indicate. There are at least 6 loci on the X chromosome and so probably about 120 autosomal loci for severe deafness. Most of these are recessive and their number can be estimated from inbreeding. In large populations the no. of loci with recessive allales for severe deafness is at least 40, whereas smaller populations have a much smaller number because of genetic drift."*

Mapping of AR deafness genes

DFNB1 - Tunisian families - 13q - affecteds are profoundly deaf or severely hearing impaired.
DFNB2 - variable age of onset - 11q13.5
DFNB3 - Bali families - pericentromeric region of 17 - profound congenital deafness

(b) Specific problems in non-syndromal deafness

AD :- Patterns described (in Gorlin) :-
(1) Congenital severe non-progressive - 60-100dB loss
Most have delayed speech
Intact vestibular system (unlike AR)
(2) Congenital low-frequency - 20-60dB
(3) Early onset progressive low-frequency (Monge)
Onset 5 - 20 years
(4) Mid frequency - 10-60 dB loss progressive begins with Mid-frequency in childhood to higher frequency later.
(5) Progressive childhood onset
(6) Progressive mixed.
(7) Unilateral
Others...

Audiogram Pattern :-

It is stated in Gorlin and by others, including Fraser that different inheritance patterns have different audiograms :- i.e. Recessive hearing loss - characteristically associated with retention of hearing in the low frequency zones.
Dominant hearing loss - audiogram is generally flat (but in Waardenburg Syndrome audiogram is like that in recessive.)

XL - some retention of hearing at all frequencies is common. (Baraitser -*"It is no means certain that this is a useful method of distinguishing one or other mode of inheritance."*

Loss is usually bilateral but can get an autosomal dominant unilateral form with reduced penetrance. Fraser states, *"the presence of unilateral or mild bilateral deafness in relatives is strongly suggestive of AD inheritance"* and it is *"easy to miss dominant inheritance especially if careful audiometry is not performed on 1st degree relatives."*

Cremers (based on a Dutch study) - *"in cases where the hearing loss in the best ear exceeds 70dB an autosomal dominant type of deafness is fairly uncommon."*

Variability :- Valerie Newton noted considerable variation in degree of hearing loss from parent to child in AD pedigrees and some families showed reduced penetrance.

AR :- Patterns described (Gorlin)

(1) Congenital severe to profound - 80-100dB loss
Some have had abnormal vestibular function...
(2) Congenital retrocochlear
(3) Congenital moderate - 30-50 dB
(4) Early onset progressive - onset 1.5 - 6 years
rapid onset - becoming profound.
(5) Progressive high frequency - onset 5-15 years

Less variability

In Valerie Newton's survey of 14 sib pairs - mean variation between sibs was only 3-4dB and no difference in the shape of the audiogram.

XLR :- Types described in Gorlin :

(1) Congenital - 7--100dB loss
(2) High frequency - non-progressive
(3) Progressive mixed with perilymphatic gusher - onset in first years of life, progressive, moderate to severe loss.

In McKusick's catalogue of single gene disorders, there are several entries but it remains to be seen whether some of these may be allelic.
i.e. (a) - with stapes fixation - Xq13-21
(b) - congenital perceptive
(c) - high freq. sensorineural
(d) - high tone neural
(e) - progressive - mapped Xq22
(f) - (deafness-hypogonadism)

Reardon reported a family not linked to XQ13-21

Mitochondrial :-

Prezant et al. (Nature genetics July 1993) identified a mutation

in a mitochondrial rRNA gene that resulted both in pedigrees susceptible to aminoglycoside ototoxicity and early-onset severe sensorineural deafness in Arab-Israeli pedigree.

ISOLATED CASES :-

Recurrence risk to siblings and offspring
- profound deafness
- less severe deafness

Cremers et al. give an overview of W. European studies and the majority give as their entry criteria the lower limit of the hearing loss. The only criterion in Fraser's study was that children be at one of the hearing impaired schools that he visited. The study designs vary considerably.

Sib recurrence risk :-

Valerie Newton found a sibling recurrence risk - overall 11% (1 in 9) BUT if >80dB loss 0.21 +/- 0.09 and if <80dB loss 0.07 +/- 0.06. This difference was not statistically significant BUT Newton postulated a 1 in 4 risk for the more severely deaf. Most other surveys give similar risks of between 9 and 12% - a full list of calculated risks has been produced by Bieber and Nance (in Gorlin).

In their genetic counselling book, Stevenson and Davidson (1976) suggest a risk of 1 in 6 based on the Stevenson and Cheeseman data from N. Ireland. The data from this study were analysed by Chung et al. who suggested that within this study population 68% were recessive, 22% dominant with high but incomplete penetrance (7% inherited and 15% new mutations), sporadic cases 9% (unrecognised infection / complex genetics) and 1% X-linked. Another study by Chung et al. based on the Clarke school for the deaf, USA (1970) suggested 27% sporadic and 73% AR (gives a recurrence risk of 18%). These higher recurrence risk figures may reflect ascertainment of more severely deaf individuals and hence a greater proportion of AR cases.

There is some evidence for non-genetic causes mixed in with the sporadic group from the Fraser survey - in his 'unknown' group, there was an excess of males and premature babies and multiple births. Also his data suggested that there might be new dominant mutations - he did not formally analyse the data for paternal age effect but noted, *"722 children in the group of unidentified causes had data on paternal age and in 71 (9.8%) the age of the father was greater than the age of the mother by 11 years or more (cf 5.1% where a perinatal or postnatal cause was identified)"*.

Offspring risk :-

Valerie Newton gave an overall risk estimate of about 6%. This is based on overall data and not broken down by severity.

OTHER ISSUES :-

(a) Marriages of deaf people - Arnos et al. comment that 90% of severely deaf adults marry another deaf person. Various possibilities can arise depending on the pattern of inheritance (if genetic) in the two partners. Various risks have been calculated in different studies. Fraser found an empiric risk of 10% of the first child being deaf where both partners were deaf and no information was available about the cause of the deafness. This is close to findings in other studies (e.g. Fay, Nance). The chances alter once a child is born to the couple, depending on whether the child is hearing or deaf. Empiric figures are given by Bieber and Nance and by Harper.

e.g. Harper :- Marriages between two deaf people :-
70% - all children have normal hearing
5 -10% - all children are deaf
20% - at least one child deaf

Parents both profoundly deaf but unrelated	1 in 10
If 1 unaffected child	1 in 20
1 affected	>1 in 2
2 affected	All

(b) Reproductive options including donor insemination could be discussed with families if appropriate.

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HEARING LOSS in a CHILD WITH LANGERHANS CELL HISTIOCYTOSIS (LCH)

Wendy Floate, Mid Cheshire Hospitals

The disorder of **"Langerhans cell Histiocytosis"** (LCH) or **'Histiocytosis X'** was first documented by Langerhans in 1868. The Histiocytosis X group of disorders were classified as Hand-Schuller Christian disease, Lettere-Siwe disease or eosinophilic granuloma of bone according to the presentation of the disease.[1]

Currently the term 'single system LCH' applies to usually bone involvement alone, or 'multi-system LCH' where several sites such as liver, lung, gut, lymph glands, spleen, bone or brain may be involved.[2]

Usually the solitary bone lesions resolve spontaneously and present in slightly older infants, children or even in adults. The multi-system form is more usually found in young infants and may be fatal.

It is a rare disease with an incidence of about 30 per year in the United Kingdom. It most frequently presents before the age of 2 years.

Normally histiocytes are formed in the bone marrow and migrate to different tissues in the body. Langerhans cell histiocytes are normally confined to the skin. In LCH there is an abnormal proliferation of these cells which migrate to other tissues where they cause damage by lysis (e.g. resorption of bone). The true aetiology is not fully known. Viral or immunological causes have been considered but refuted. Current research suggests tumour clonality[1] which may be benign but possible malignant development has not as yet been fully investigated.

The most commonly affected sites are limb or skull bones presenting often with limb pain and later diabetes insipidus where the pituitary stalk is affected following lysis of skull bone. Multi-system disease has various presentations but usually in an ill young infant.

Chronically discharging ears due to necrosis of mid-ear ossicles seems to be reported in standard textbooks. Personal communication with the 'Histiocyte Society', Hammersmith Hospital, indicated that deafness due to inner ear destruction from temporal bone lysis has been reported with deafness remaining after remission of the disease, but documentation of incidence and type of hearing loss is difficult to find.

Locally we have a young boy with acquired severe mixed hearing loss following development of LCH of limb and skull bones. This boy passed early hearing screens and had normal language development. He was using sentences clearly before the age of 2.5 years.

He presented at the age of 2.5 with a limp in the left leg and was referred by his GP to an orthopaedic consultant. An osteolytic lesion of the left iliac bone was found. Needle biopsy confirmed presence of LCH. He was treated with Indimethacin for bone inflammation and pain by a paediatric orthopaedic consultant.

Mother noticed hearing difficulties at the age of nearly 3 years and was recommended by the orthopaedic consultant to see her GP who referred him to an ENT consultant. He had EUA's and a grommet was inserted in the left ear since glue was found; the right ear was said to have discharge from otitis externa, so was packed with gauze soaked in gentamycin.

He developed polyuria and thirst and was only referred to the local paediatrician at over 4 years of age. Diabetes insipidus was diagnosed and treatment with vasopressin was started. Mother expressed concern that his hearing was deteriorating despite ENT treatment, and in the last 6 months his speech was disintegrating till language seemed to have disappeared.

At the same time he was nearing school admission and was referred to the educational psychologist who referred him to the advisory teacher of the deaf. She reports that he was impossible to test.

The paediatrician referred him to the SCMO in audiology as a case of extreme parental concern at the age of 4.5 years. It was quite apparent that he was a frightened, withdrawn little boy who was not able to understand speech. Possibly, he was also having skull bone pain.

He would not accept a bone vibrator nor earphones and could not be conditioned to perform to sound at the age of 4.5 years.

He was diagnosed by the distraction test with warbler

held near his ear as having a bilateral profound flat mixed loss averaging >95dBA in the 5 frequency range of 250Hz-4000Hz. He had a normal tympanogram on the right but debris and discharge in the left canal so tympanometry was not done on that side.

High gain extended low frequency post aural amplification was provided promptly. He was then admitted to a partially hearing unit. We are pleased to report that when last seen at the age of 5 years and 10 months he was using sentences again and was learning to read a few words.

His hearing on a recent audiogram shows a mixed severe loss with slightly better levels on the left side. He was still not ready to do a full audiogram. He continues to have discharge and debris in both ear canals, but will not go without his hearing aids.

At the time of diagnosis of his hearing loss MRI scans revealed very extensive destruction of the right temporal bone and the pituitary stalk was affected. Treatment from a paediatric oncologist and the local paediatrician was started following the scan at the age of 4.5 years. The cytotoxic drug Vinblastin was given regularly through an indwelling portha-cath, plus prednisolone for nearly one year until his condition improved.

He is no longer receiving indomethacin and the vinblastin and prednisolone have been stopped. Recent MRI scans done 9 months ago show no lytic lesion in the left iliac bone, slight improvement in the right temporal bone but the pituitary stalk is still affected. Thus there is radiological evidence as to cause of loss on the right. Specialised CT scan of the petrous temporal bones was not done. Clinically discharge has occurred on both sides; therefore, clinically active disease of the left middle and maybe inner ear seems to have occurred. Both ears have been treated with gentamycin, possibly for a few months, but we do not know if this contributed to hearing impairment.

The active disease seems to have remitted but he is left with diabetes insipidus, still needing vasopressin, and a mixed severe hearing loss.

Audiometry results (age 5 years 10 months)
(at the time of remission)

Air Conduction

	5ooHz	1KHz	2KHx	4KHz
Right ear	80dB	115dB	85dB	95dB
Left ear	60dB	100dB		70dB

Bone conduction 60dB 70dB
(Masking not possible)

Permission was obtained from parents and paediatrician to use information from medical notes.

References

1. *Cotter F., and Pritchard J., BMJ, 6972, vol.310, 14/1/95 Clonality and LCH*
2. *Langerhans Cell Histiocytosis (a parent booklet)*

FUNCTIONAL HEARING LOSS
Information and advice for School Medical Officers and School Nurses.

by LINDA WALLIS

The term 'functional hearing loss' (FHL) is one of many used in the literature to describe any hearing loss which cannot be accounted for by an organic cause.(*1)

FHL is usually diagnosed following discrepancies between results of difference hearing tests and the patient's response to sound in non-test situations. It may be superimposed on an organic deficit.(*2) (D.Y.Aplin and V.J.Rowson, 24th April 1989)[1]

My Survey.

I have been an audiology sister working in the community for 8 years - and previously a School Nurse for 3 years. I needed to know more about functional hearing loss and the problems which the afflicted children were experiencing. My colleagues - School Doctors and School Nurses - have also expressed a wish to be better informed, so I decided to collate my findings in this survey.

My parameters are:
1) Children tested by me.
2) A two year period - January 1992 - December 1993
3) Children at School, i.e. school aged 5 - 18.
4) Children with persistent test failures.

It is important to note that I had access to social, emotional and health histories in order to complete this survey. However, many audiometricians and ENT departments will not have access to this information and therefore can make only a limited assessment
.
Further relevant information should be given when the children are referred because of failed hearing tests, so that what is really wrong with them can be managed effectively.

Early Findings

As early as 1959 Dixon and Newby[3] talked of children with a functional - or non-organic hearing loss - as:

> *Children with a difficulty working with one teacher in particular, or where there is conflict in the home.*
> *Children where there is pressure to do well, who are in need of sympathy from parents, peers or teachers.*
> *Children who use hearing loss as an excuse for poor academic work.*

They also noted occasions where hearing difficulties were not expected by parents or teachers because of adequate hearing and clear speech of the child, and were therefore surprised by the failure of hearing tests in school.

Aplin and Rowson[1] came to feel that boys, when maladjusted, over reacted aggressively; girls adopted withdrawn behaviour such as FHL. Although this is a generalisation, I did find 28 girls and 13 boys in my survey.

Categories

A (i) The child who can hear but does not respond until confident.
(ii) The child who is "playing up", who does not want to be doing the test.
(iii) The child who wants to fail ?attention seeking? Why?

B (i) Children who have difficulties processing sound.
(ii) Children who are emotionally disturbed.
(iii) Children with perceptual difficulties.

Children with central deafness (or congenital auditory imperception) which is rare in its severe form, but in a lesser degree may be associated with delayed language development and other learning difficulties. (Dr. Isobel Price[2])

Breakdown of Categories

Category A

Children who have behavioural problems.

i) Shy
Slow movers.
Learning difficulties.
Withdrawn.
Anxious about being wrong

ii) Jokey - tricksters - children who treat the test as a joke.
Show offs (acting for peer group)
Reluctant (wants to be doing something else).
Naughty (behaviour that interferes with testing).
Wants to be promised a reward.

iii) Not doing well in school, needs a reason for this.
Has been in trouble for "not listening".
Likes to have family or friends concerned seeks attention.
Wants to be "different".
Tired.

(Examples given by Dr. Price[2] from her experience.)

Category B

Children who have conditions which prevent them from responding to sound normally, in spite of normal middle ears, inner ears and the VIIIth cranial nerve (Auditory).

i) Children who have psychotic or autistic features, whose withdrawal distorts their response to environmental stimuli, including sound. They fear what they do not understand, and hence are often fearful of the test situation, machines, etc.

ii) Children who are emotionally disturbed
Neglected.
Abused.
Latent anger (like elective mutes).
Secretive.
Guarded.
Under pressure.

iii) Unable to make sense of sounds.
Unable to make sense of words.
Mental handicap - no response.
No interest in test sounds.
Little comprehension of test requirements.

(Examples given by Dr. Price[2] from her experience.)

Breakdown of Survey findings

Sex	Boys 13	Girls 28
Ages	6 years -1	12 years -7
	7 years -5	13 years -2
	8 years - 4	14 years -1
	9 years -5	15 years -4
	10 years -5	16 years -2
	11 years -5	

Reasons

Special educational needs identified - school test not ability appropriate 2
No known reason 2
Too frequent testing 3
Abuse 14
Neglect 5
Bullying at school 3
Pressure at home - to do well/ emotional families/ jealousy 7
Other illness / asthma 3
Shy - lacking confidence 3
Total 42

1 child in 2 categories (Too frequent testing - identified as more than 2 times by School Nursing Service before referring to clinic - protocol, test x 2 then refer to clinic).

Considerations in FHL

If someone presents with a functional hearing loss the following factors should be considered:

The child identifying with other family members who are deaf or otherwise disabled.
Children from broken homes.
Where there is parental conflict with each other.
The intelligence / ability of the child.
The frequency of testing.
The likelihood of sexual or physical abuse.
If there is neglect, or poor quality parenting.
If the child is bullying or being bullied in school.
Is the child ridiculed for any reason?
Is there pressure from parents, staff or even the

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child, to do well in school?
Is the child jealous at all?
Are they able to discuss problems with parents, school staff or friends?
Is there illness of any kind, child feeling unwell often? i.e. Many colds, Asthma, Eczema, Physical handicap causing tiredness.
Shy, lonely, unable to make friends easily, or lacking in confidence or self esteem.
The child who is lacking in self esteem may be trying too hard to be accepted by others.
Is the child tired most of the time, late bed time, watching TV or videos for too long?
Is the child a poor sleeper, worrying about something or having a cramped sleeping area?
Does the child fear failure in any way?
Is there a background of ENT attendances or glue ear and is there already knowledge of audiometry testing?

If you think someone has a functional hearing loss.

Children who are thought to have a FHL must be seen in audiology clinic, but it helps us with diagnosis and management if we have as much information as possible.

Please record:

Why the test in school was carried out - routine or because of concern?
Does the child him/herself complain of hearing difficulty?
The teacher's and parent's view of the child's hearing ability.
Information re: the child's academic progress.
Whether there are any social / emotional difficulties known.

Your assessment:

Were the child's responses on audiometry consistent or erratic?
Record the child's responses to conversational and/ or quiet speech, spoken from behind or with mouth covered. (A child with a true 50dB loss cannot hear what you are saying without lip-reading.)

In clinic:

Various tests may be used to clarify the audiogram results:

1 Speech discrimination tests (Word lists)
Speech audiometry (Word lists via headphones).

2. Pointing to ears:

Make 20 or 25dB signals and ask child to indicate "which ear?". Wait a while before making a signal and count as correct if child indicates, even if it is the wrong ear.

3. Say "Yes":

Once again, make infrequent signals, with a sizeable waiting time. Count as correct if a child says "no" at appropriate time, or "I didn't hear that".

4. Counting beeps:

Say you are going to give beeps or signals from 1 to 4 and ask the child to count and say how many (1,2,3,etc.).

Once interested, turn signal down to under the decibel level indicated on audiogram. Actual decibels or Kilo Hertz do not matter for this test. It just indicates that hearing is better than previously indicated.

5. Increase, then repeat volume.

This is a quick test. By nature it cannot be prolonged, but it helps to confirm findings.
If a result of 40-50dB is given, but hearing is thought to be better than that, try the following test:

Give a signal at 1,000Hz at 10dB, then 15dB, then 20dB, then 20dB again. If this is done quickly the child often responds on the second or third 20dB level, thinking it to be progressively louder.

Outcome

On reading round the subject of FHL I found several articles, all by Audiologists and Audioscientists, relating to the hearing side of FHL and how to disprove the findings of Audiometry. I wanted to know more about the outcome, once the child is no longer c/o ENT or Audiology, once it's proved there is no hearing loss, and the "crutch" of a hearing loss problem is removed.

The "real" problem is still there; the child still needs help.

When I started my survey, I could find no mention of child abuse as a factor, but there is now increasing evidence of this.

Much counselling is needed, and as it is school children (not under 5 years old) who have FHL, I feel it is the School Nurses or School Doctors who are in the best position to help, with support from teachers, parents, Social services, Educational Psychologist and Child Psychiatrists - as appropriate in each case. If associated with child abuse, the Key Worker should co-ordinate counselling or referral made to Social Services.

When it's not FHL

I have found no evidence of FHL in children under 6 years during the two years of study. If a child cannot do a test because of not understanding what is required, it cannot be called FHL. A more appropriate test should be done.

Similarly with children who have Severe Learning Difficulties, the School Nurses and School Doctors should refer to Audiology Clinic for an ability appropriate test. In Mid Downs Health Authority area these tests are done in school by the Audiology Sister and School Nurse, with guidance and the expertise of the Audiology SCMO when required.

Conclusion

In most cases, the children had once been in the care of Audio Clinics or ENT department or had been shown concern or sympathy by the School Nurse, which meant receiving individual attention by a caring adult. Some parents comment on catarrhal problems in the past when, as toddlers, the children were taken to clinic, but the catarrhal problems have long since cleared up. In other words the children knew about hearing tests and what was required of them.

It seems significant that abuse, emotional, physical and sexual, and bullying may predispose to FHL, especially in girls.

In most cases the children who come into category A should be able to be identified and addressed by the School Nursing Service. Children in category B may need the more detailed testing of the Audiology Service.

Questions to consider

I hope the information given will be useful in alerting professionals to what the children are '*not saying*'.

Why do children choose not to hear?
What is it they want to avoid hearing?
Why do they need the concern of the carer, if they don't have a hearing loss?
What is the real problem?
Do we make things worse by constant testing?

Hearing Classification

Printed in a paper by Dr. M. Nolan - 1.2.80 (iv) - additions by Linda Wallis

Normal hearing:

Hearing levels at or better than 15dB bilaterally for frequencies of 250Hz to 4,000Hz.
(If testing in school 20dB is acceptable) LW
Middle ear function normal (no catarrhal problems) LW

Speech discrimination consistent with normal hearing. (It takes approx. 40dB to hear a word correctly although a beep can be heard at 15dB). LW

Child orientated tests consistent with normal hearing.

Conductive hearing loss:

Depressed air conduction thresholds, normal bone conduction thresholds. Signal made to bone over cochlea, just behind ear). LW

Abnormal middle ear function (tested by tympanometry). LW

Speech discrimination scores consistent with audiogram performances on child oriented tests consistent with audiogram.

Bilateral Sensori-Neural Hearing Loss

Depressed air and bone conduction thresholds, normal middle ear function. Speech discrimination

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consistent with audiogram. Performance on child orientated tests consistent with audiogram.

Unilateral Sensori-Neural Hearing Loss

Normal air and bone conduction threshold one ear, depressed air and bone conduction for the other ear. (If the hearing loss is severe in one ear, a "shadow" result can be obtained in that ear, because sound is eventually heard by the good ear.) LW

Mixed Hearing Loss LW

When a child has a sensori-neural loss, made worse temporarily by a conductive loss. LW

Functional Loss

Inconsistencies between tests as described in the text.

References

1. D.Y.Aplin & V.J. Rowson, Centre for Audiology, Education for the Deaf and Speech Pathology, The University, Oxford Rd, Manchester.

2. Dr. Isobel Price MB ChB MFCM DObst RCOG DCH SCMO Audiology, Mid Downs Health Authority.

3. Dixon & Newby 1959, Children with 'non-organic' hearing problems. Archives of Otolaryngology, RNID. library, 3300332 Grays Inn Road WC1X 8EE

4.Dr. M. Nolan, Department of Audiology and Education for the Deaf, University of Manchester, M13 9PL

Further details of 'Central deafness' can be found in 'Clinics in Developmental Medicine - The Child Who Does Not Talk. No.13' - Neil Gordon, P62-64 and Ian Taylor P65-68

Appendix

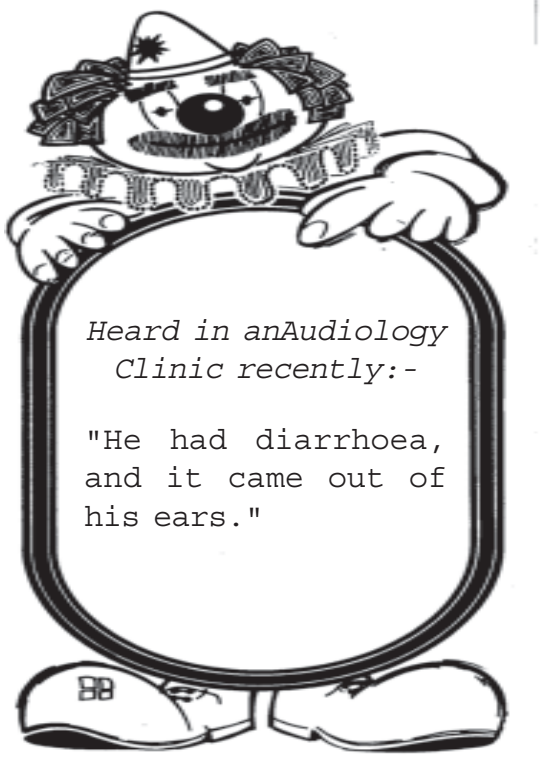
Children in Survey

1. Boy	aged 7	Suffering from Asthma, shy and nervous.
2. Girl	aged 7	Abused at home - father.
3. Girl	aged 12	Slight learning difficulty - late diagnosis.
4. Girl	aged 16	Abused at home - now in care.
5. Boy	aged 9	Difficulties at home - previous Audio problem, new baby at home ? jealous.
6. Girl	aged 11	Abused at home - bullying at school.
7. Boy	aged 8	Family difficulties - immature mum - family aid - vulnerable child - dominant grandmother.
8. Girl	aged 12	Sexual abuse - mature for 12 - dad in prison.
9. Boy	aged 10	Abused sexual and physical - at home.
10. Girl	aged 8	No clear reason indicated.
11. Girl	aged 12	Mother seriously ill - moved house recently - new baby, mother overanxious (noted by Health Visitor).
12. Girl	aged 15	Father alcohol problem - sexual abuse by father's friend.
13. Boy	aged 15	Sexual obsession at 12 - fostered/adopted - behaviour problems.
14. Girl	aged 8	Bullied/harassed by one teacher.
15. Girl	aged 11	Asian - shy ++.
16. Girl	aged 12	Poor attender at clinic - hyperactive - dislocated shoulder.
17. Girl	aged 10	Sister with severe learning difficulties - has allergies herself and lacks confidence.
18. Girl	aged 10	Asian (moderate learning difficulties), late diagnosis.
19. Girl	aged 7	Mother sure there is a problem - not reassured by results - school cannot reassure her either.
20. Boy	aged 12	Overweight - Asthma allergies
21. Girl	aged 7	Emotional difficulties at home - mature for age - naughty in class since dad left.
22. Boy	aged 11	In care of Social Services - expelled from all schools attended.
23. Girl	aged 16	Pressure from parents to do well - colic pains - cries++, small rectal tear.
24. Girl	aged 10	Medical, learning and social emotional difficulties - c/o Educational Psychologist
25. Girl	aged 11	Bullied at School
26. Girl	aged 15	Depressed mother - child is over fond of new stepfather.
27. Boy	aged 7	Parents had difficult divorce - several house moves - boy very worried about past and future - moving abroad - soon new marriage and in-laws.

28. Girl	aged 15	Emotional attachment to mum - damage to and jealous of new baby. Social Services involved, poor concentration in school.
29. Boy	aged 12	Poor care as a baby - operation at 2 weeks old - not doing well in school - changes of address++ - changes of school.
30. Girl	aged 11	Slight sensori-neural loss Rt ear, but OK - poor attender at Clinic - every test in school showed a severe loss, but for speech, normal hearing.
31. Boy	aged 10	Mother with new man - Social Services involved - family down-market since leaving father. Quick changes of school unsettling.
32. Girl	aged 13	Sexual abuse - lodger - sexual abuse in family - other members.
33. Girl	aged 12	No obvious reason - too frequent testing.
34. Boy	aged 8	Father left home - he was deaf c/o tummy aches - needed paediatrician and psychotherapy - sees father now.
35. Boy	aged 9	Poor parenting - failure to thrive, accidents/casualty++ drug abuse parents.
36. Boy	aged 9	Headaches / vomiting / migraine - too frequent testing and referral to ENT - always satisfactory.
37. Boy	aged 6	Letterwork in school poor - cries a lot to get mother's attention - cries ++ in school to avoid trouble - jealous of siblings.
38. Girl	aged 13	No known reason for FHL.
39. Girl	aged 14	Sexual abuse - father - violence to child and mother - child and sister are both in care.
40. Girl	aged 9	Mother c/o psychiatrist - very immature - disruptions in home life - frequent moves.

Functional Hearing Loss
By Linda Wallis, RGN CMB Part 1, NNEB, School Nurse Cert. Audiology Sister, Mid Downs Health Authority, Crawley Horsham NHS Trust, Mid Sussex NHS Trust. 7th Feb. 1994.

I would like to express my thanks to Dr. Isobel Price MB ChB MFCM DObst RCOG DCH for her help and support in preparing this survey. Also to Sue Jeal RGN SCM HV MA, my Nurse Manager



For further information, help or advice:-
Telephone 0121-443 5777 or Fax: 0121-443 5999
or write to:-
20 Branscombe Close, Kings Heath, Birmingham, B14 6PP

MSc Funding. The Saga

Five years ago I was a CMO on the Doctors' retainer scheme happily fitting in two sessions a week for school health with family commitments. Our new CCP started rocking the boat by telling us that if we did not specialise and pull our socks up we were doomed to be overrun by GPs. I very half-heartedly volunteered an interest in Audiology but tried to keep myself in the background. When my husband took voluntary redundancy and went into teaching I took up a 50% contract and found myself on a working party trying to improve the run down Audiology service in our area. It became clear that I was going to have to change my attitude and take some initiative in furthering the service. While on Holiday with my sister in Oxford I decided to try and contact the SCMO for Audiology there and see how their service was run.

My Phone call was returned at 8am(!) the following morning and Deirin Nicholls invited me to come and see her there and then. So I jumped on my bike and cycled round to the John Radcliffe and within a couple of hours my whole perspective on Audiology had changed and I was determined to find out how other people ran their service. I made contact with people in other health districts and began to realise that our Audiology service was like something in the dark ages! Everyone I approached (all BACDA members) went out of their way to be helpful and encouraged me to join BACDA.

When I returned from the summer holiday that year my CCP didn't know what had hit him! I was as keen as mustard and brandishing the BAAP Policy document that he had given me pointing out that it quite clearly states that the lead doctor should have the MSc in Audiological Medicine. He gamely took a deep breath and told me to go for it, that he would back me all the way but that he couldn't promise that the funding would work out. He was as good as his word and wrote to the administrators and the commissioners telling them why they should fund me, presenting a strong case for the need to improve the Audiology service. Meetings were held with unit administrators and general managers and they did not seem to be able to see their way to either paying the course fee or my salary on study leave. I wrote to various charities for the deaf and suppliers of Audiological equipment (one of whom actually replied to my cheeky request) and even applied to the South East Thames Regional Authority for a grant through their Research and Development Scheme. When they turned down my application at the end of July (by which time I had applied for and been given a place on the ILO MSc course) another desperate meeting with the managers took place and someone suggested asking the local postgraduate dean if he had any funds for this type of course. We all knew what his answer would be but as an afterthought he suggested I try the British Postgraduate Medical Federation. I wrote to them asking for advice as to whom I could approach for funding and on my return from the summer holiday in late August found a letter from them saying they would be happy to pay my course fees up to £2000 if the Trust would pay my salary while I was on the course. During the same month various other funds had become available to the Child Health Unit and they could now see their way to covering my work and supporting me. By this time I had worked several extra sessions, partly as a good will gesture, but partly because I knew they would pay me for work already done and I was so determined to do the course I was even considering going on unpaid study leave.

So take heart if you are considering doing the MSc. It is well worth all the effort of getting the funding even though they let you go right to the brink before deciding. It is also well worth the effort and toil of completing the course but be warned - it is not a course for the faint hearted - you have to be really single-minded and work hard and tell all your friends to write you off for one year (or two) while you are studying. Like many others, I have survived to tell the tale and hope my Audiology service will reap the benefit.

Ceridwen Deacon
CMO in Audiology

The "Can your baby hear you?" form copyright McCormick 1981

Hints for Parents

Can your baby hear you?

Here is a checklist of some of the general signs you can look for in your baby's first year:

YES/NO

Shortly after birth
Your baby should be startled by a sudden loud noise such as a hand clap or a door slamming and should blink or open his eyes widely to such sounds. ☐☐

By 1 Month
Your baby should be beginning to notice sudden prolonged sounds like the noise of a vacuum cleaner and she should pause and listen to them when they begin. ☐☐

By 4 Months
He should quieten or smile to the sound of your voice even when he cannot see you. He may also turn his head or eyes toward you if you come up from behind and speak to him from the side. ☐☐

By 7 Months
She should turn immediately to *your* voice across the room or to very quiet noises made on each side if she is not too occupied with other things. ☐☐

By 9 Months
He should listen attentively to familiar everyday sounds and search for very quiet sounds made out of sight. He should also show pleasure in babbling loudly and tunefully. ☐☐

By 12 Months
She should show some response to her own name and to other familiar words. She may also respond when you say 'no' and 'bye bye' even when she cannot see any accompanying gesture. ☐☐

Your health visitor will perform a routine hearing screening test on your baby between six and eight months of age. She will be able to help and advise you at any time before or after this test if you are concerned about your baby and his development. If you suspect that your baby is not hearing normally, either because you cannot answer yes to the items above or for some other reason, then seek advice from your health visitor.

Produced by Dr. Barry McCormick
Children's Hearing Assessment Centre, Nottingham
Printed by The Sherwood Press (Nottingham) Limited

The situation regarding its availability.

This form is available directly from the J & B McCormick partnership in an A4 or A5 (pre-punched parent held record) format. Anybody who prints or photocopies the form without the author's permission is violating the copy right law. Infringements are currently being investigated and the reader is asked to check if the version being used in their authority have been produced with the author's permission.

The History

The author produced this form in 1981 and published it in 1988 after extensive field trials in the Nottinghamshire Area Health Authority. The author had given permission for his local health authority to print the form for local distribution and this was included in the local supplies system for stationery and printing. Following the receipt of a number of requests from other health authorities to obtain supplies of the form the author asked his local authority if a supply arrangement could be set up. The reply was "NO" because of the administrative inconvenience and complications involved in organising a supply and invoicing procedure.

In order to satisfy the demands the author arranged fro the form to be printed and supplied through the J & B McCormick partnership for which he holds a 5% intellectual rights share. This partnership has been set up to satisfy a demand for a service and was dependent upon orders to maintain its viability. Clearly any unauthorised printing and copying of the form would not only breach copyright but would undermine the viability of the very service that had been set up to satisfy demands.

With the introduction of parent held record schemes, a number of authorities (and printers) have taken it upon themselves to reproduce the "Can your baby hear you?" form but exclude acknowledgment to the author and others are of a very poor quality with changed wording and wrong attributions to the author