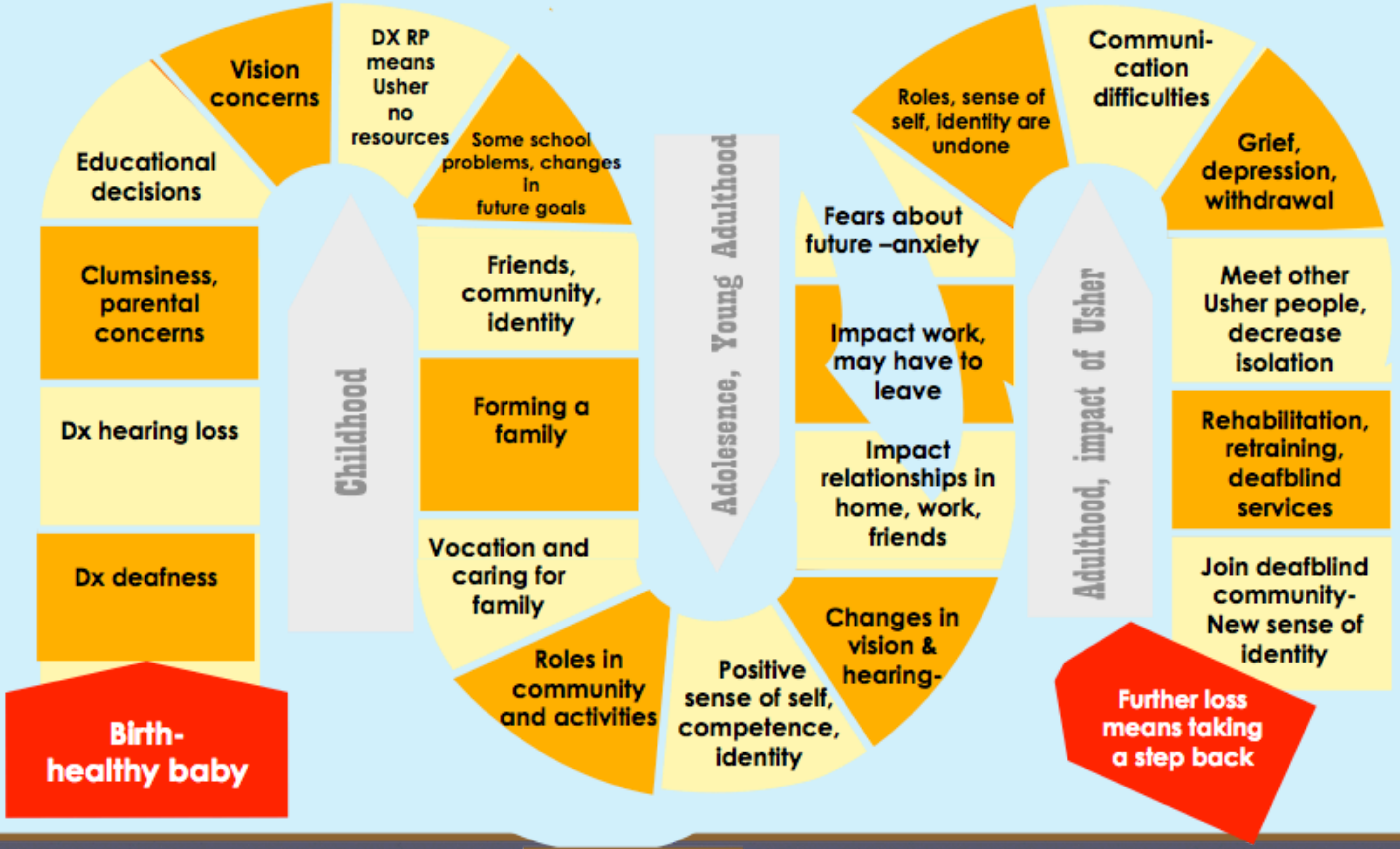
Diagnosis of Usher Syndrome

Information and and Suggestions

This is true

- if you work with Usher children, you must know Usher adults
- If you work with Usher adults, you must find out about their lives as children.
- -Sarah Gardner, a psychiatrist in Sweden

An Usher Life



Birth- healthy baby

Dx deafness

Dx hearing loss

Clumsiness, parental concerns

Educational decisions

Vision concerns

DX RP means Usher no resources

Some school problems, changes in future goals

Childhood

Friends, community, identity

Forming a family

Vocation and caring for family

Roles in community and activities

Adolescence, Young Adulthood

Positive sense of self, competence, identity

Changes in vision & hearing-

Impact relationships in home, work, friends

Impact work, may have to leave

Fears about future –anxiety

Roles, sense of self, identity are undone

Communi-cation difficulties

Grief, depression, withdrawal

Adulthood, impact of Usher

Meet other Usher people, decrease isolation

Rehabilitation, retraining, deafblind services

Join deafblind community- New sense of identity

Further loss means taking a step back

An Usher Life

Who am I?

- Clinical social worker
- Involved with deaf, hard of hearing & folks with Usher for 34 yrs as a professional & as just me
- I’ve written and published some, and presented in Denmark, Netherlands, Spain, Belgium, England, and the US
- 20 years of work in a major medical center in Pediatrics
- Less tactile sign now because my shoulders have betrayed me
- Still get phone calls from newly diagnosed families
- I have an adult son with an acquired disability- and I, like every parent here, am a Class- A worrier

A word about Mary Guest

- Mary, who died last year, was the Mark Dunning of Usher in the UK- before there was an internet to connect people.
- She worked with SENSE, fought for funding, brought families and people with Usher together around the UK and indeed around the world.
- Her accomplishments are too numerous to list- and I was privileged to call her friend and colleague.

Why this presentation?

- At the last conference, the issue of what to tell families came up during informal discussion
- With Mark Dunning and the Coalition and friends, I had a group of families we could ask about these issues
- We asked parents and people with Usher
- I sent a survey out through the coalition and also by sending the document to people I know
- Thanks to Elias Kabakov in Israel who did the work of getting into Google- I'm still old school!
- And to every person with Usher and family who took the time to answer

A message from the mom of a 21 year old son with Usher

- “I hope that you'll encourage parents to realize that their child who has Usher Syndrome has a world of possibilities ahead of them as long as they keep the bar high, learn the necessary skills and believe in their children. ”
- This young man is in college and working now
- They had a tough time with diagnosis because when he was found to have vision and hearing problems, his gene had not yet been identified - he was incorrectly diagnosed

Survey: I asked:

- about their child or themselves, diagnosis, current age, age at diagnosis
- about who gave them the diagnosis and how empathic they were
- Whether or not they had seen a geneticist
- what they think could have been better and what information do they have now that they wish they had then
- What message they had for their providers

Who responded?

- 39 surveys were returned, 37 from the US, one from the UK, one from Canada and one from Denmark, and states from NY to Alaska
- Quite the mix: parents of young children, adults with Usher, parents of adults with Usher, and two parents of adults with Usher and that Usher adult together. Three families have 2 children with Usher
- It’s not so easy to analyze these by age because the time frames of diagnosis and ages of respondents are so varied.
- There were adults diagnosed 30+ years ago and parents whose child was diagnosed within the year. But there was consistency in what they reported.

Reactions to diagnosis

- Shock, devastation, confusion, despair, guilt
- Traumatic, left feeling hopeless, helpless not knowing where to turn
- Future is unkown, plans gone
- Can’t cope
- Too much information and not enough
- Depression and anxiety

Comments about diagnosis meeting

- Too much info given too quickly to process
- Too little practical information
- Take them home and teach them Braille
- Doc excited about finding his first Usher case, not our reaction
- Gave me reading material and web sites
- Gave me no contact information for real people
- Too clinical and technical
- Message was – there is no hope and nothing to do

Comments about empathy- parents reported:

- their providers were empathic and patient; but some said that their doc had no empathy and no patience
- Some reported that their doc seemed more interested in getting their child into a research study
- Some reported that their doc just made no acknowledgement of the devastation wreaked upon the family in that moment
- “The doctor offered no hope; we need to be told there is hope”

Empathy

- Of the 37 respondents, 26 answered this question
- 9 people rated their provider 1,2,3. And 7 rated their provider as a 1.
- 10 people rated their provider 8,9, or 10. 3of these ratings were a 10

Genetics

- 39 responses representing close to 50 children
- Only 15 families were referred to geneticist
- 17 families stated they were not referred
- 7 people gave no answer
- This was NOT completely a situation of the older ones not being referred and the younger ones being referred.
- There were older ones referred and younger ones not,

Suggestions from parents

- Prepare for meeting. Give written material- we should not have to search for it
- Connect us with families with Usher, including Coalition
- Realize we will ask the same questions again and again because we can't absorb everything in one meeting.
- Call us in a week or have a non physician call to see how we are doing
- Be patient and empathic: imagine if your world was devastated in a minute in someone's office
- You may have given this info a million times, but we are hearing it fort the first and only time.

Suggestions from literature

- So, I looked on Medline for articles on Parental reactions to diagnosis of Usher Syndrome, and guess what I found?: ZERO

- There were articles on diabetes, irritable bowel, Down Syndrome, Intellectual disability, heart disease, and even one on vision impairment

Two models

- Community Linkage Team- at Great Ormand Street Hospital- UK- a team -One member assigned to family on day 1, reviews med info, is contact person, connects family to groups, other families, social services, school reports, social support, facilitates access to programs. Team is comprised of skilled NON-medical professionals
- Deaf Blind consultants- Denmark- a children's deafblind consultant assigned to every family where there is a person with a deafblind diagnosis including Usher. Provides support & counseling to the family.

Grief, Depression, Anxiety

- Grief is not linear
- Grief, depression, anxiety is not once
- Grief, depression, anxiety are cyclical and recurrent
- Grief, depression, anxiety for both the person with Usher and the family

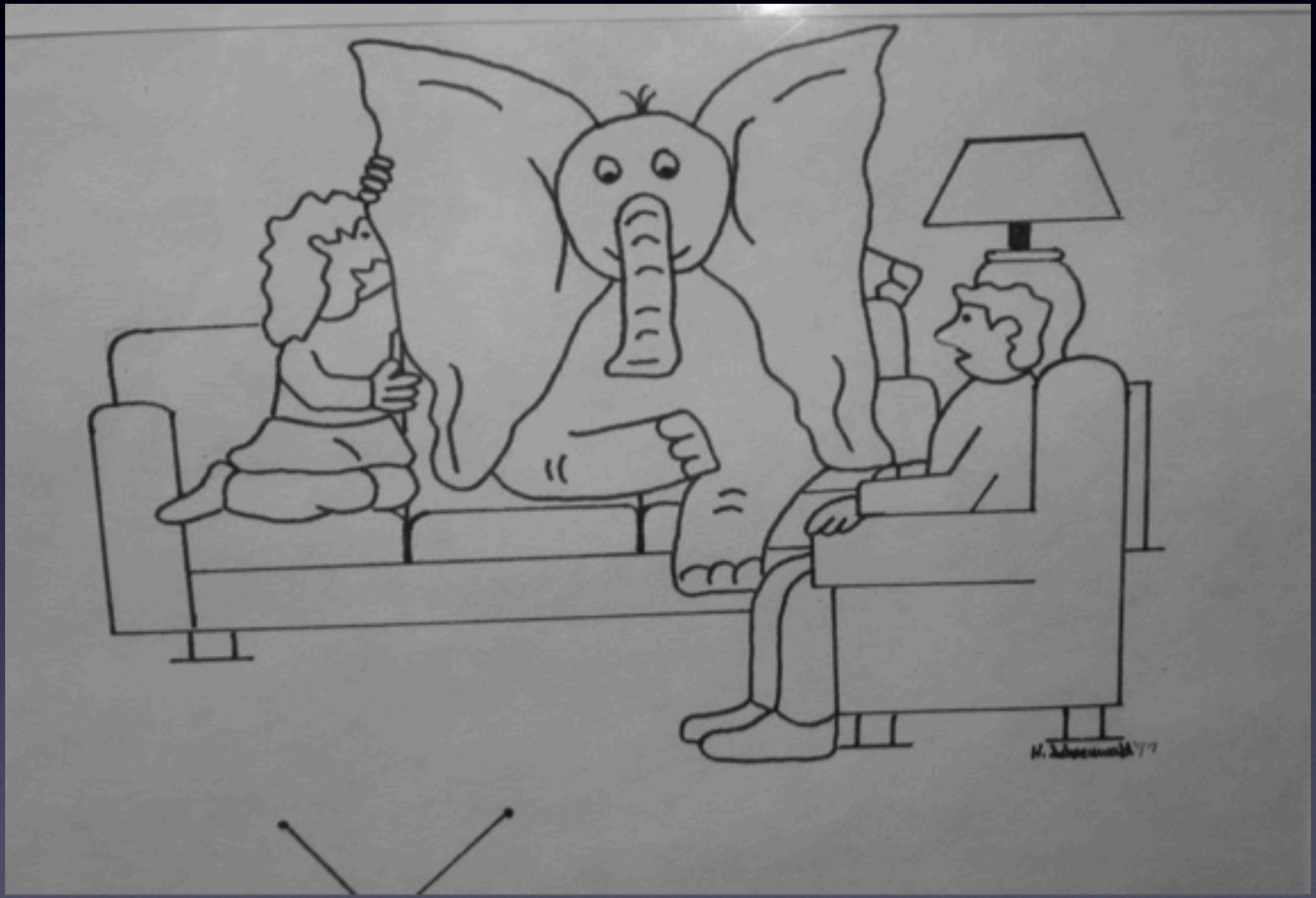
Resurgence of Emotional Issues:

Person with Usher and & Family

- When there is further vision or hearing loss
- When normal developmental events don’t happen on time
- When there are unrelated losses or major life events/ stages
- When new accomodations have to be made

The elephant in the living room

This was drawn by a young woman with Usher II after we talked about her family who never discussed Usher



From the survey: Parent concerns

- What and when should I tell him/her
- This is all my fault...
- What happens as my child with Usher get older?
- Will my child be independent? Drive? Go to college? Work? Marry? Have children?
- Should they limit their career choices?
- What will happen when I am gone?

Parent Responses

- Depression and Anxiety, which Mark has written about so eloquently
- Dealing with these requires time, perhaps some help, some action, contact with other families and with people with Usher
- As one professional with Usher wrote, “I had a psychologist who didn’t know about Usher, but that person knew about loss”

Coping Strategies- briefly

- The literature talks about coping strategies as if there are only two categories: Problem-focused coping and emotion focused coping
- But really it's a combination of the two that gets us through the day
- For families and for folks with Usher, it's about learning, information, services, and meeting others that leads to sense of competence and mastery, along with professional emotional support, if needed

Talking about Usher- family

- Children & teens have to be able to ask parents– Be the askable parent
- Talk about it a little, a lot- Mark
- Normalize problems- taking a hand in the dark, introducing others with Usher

talking about Usher- family

- do NOT say “you are going blind”- this is not useful- because for any one person, no one knows
- Be prepared to explain things <u>many times</u>- kids understand differently at different developmental stages- <u>discuss a little bit, but often</u>
- Build in contact with slightly older Usher kids and successful adults

Services

- In some places, available; in other places, not
- Teens cannot feel good about themselves is they have no skills to master what is happening to them and in their environment,
- nor can they imagine a future if they have never met a successful adult with Usher.
- Early introduction of O & M and Braille, even if it is presented as something that may never be needed
- Counseling is effective if the other skills are being taught..

Denial

- Much maligned but it allows hope
- Purpose is to protect the integrity of the person
- Unconscious mechanism
- Functional vs dysfunctional denial
- If it's functional, support it- "I will learn these skills even though I will never lose my vision

The skills no one wants to talk about

These are signs of independence, not dependence

a comment on mental distress

- Everyone needs help sometimes, parents too
- Usher Syndrome does NOT cause mental illness
- Usher Syndrome doesn't prevent mental illness
- Some teens, adults, families can benefit from good mental health treatment in times of stress and loss
- Sadly, people with disabilities often get poor mental health services

some things for professionals NOT to say

- “I understand”
- “You will never go blind”
- “you will go blind and you have to accept this”
- “Don’t worry”

In summary

- Usher is about loss, change, adaptation, information, communication, normalization, and fighting against the isolation that can be Usher
- Our roles are about how to mitigate the impact on those we teach and treat, are friends with, and love