



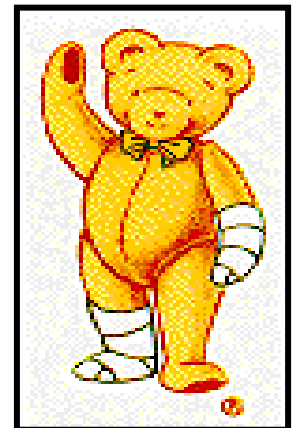
Helping families affected by 'rare diseases'

Lisa Smith

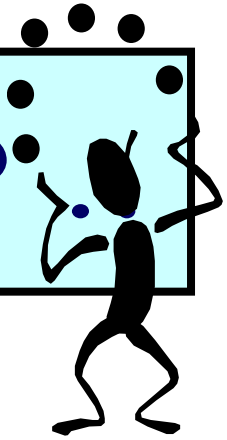
Genetic Counsellor

Department of Clinical Genetics

The Children's Hospital at Westmead



What is a genetic counsellor?



Always work as part of a tertiary service

Role:

- Allied Health -
 - Work with a consultant - usually a geneticist
 - Work in a multidisciplinary team with consultants of other specialties and therapists
 - Liaise with other support systems e.g. GP, local therapists, schools etc
- Education - e.g. genetics, genetic testing, reproductive planning options
- Facilitate decision making + practical help and referral
- psychosocial counselling support for all family members

What are rare conditions ?

- * Conditions that 'no one' has heard of..
- * Conditions that 'no one' wants to look at..
- * Conditions that isolate..
- * Conditions that no one wants to think can happen to them..
- * Conditions that no one can fix, or make better..

- * Conditions that challenge, confront, repel ..

- * Conditions that cause tragedy..

- * Conditions that lead to a 'bottomless pit of need'
- * Conditions that change people

What 'help' do we offer families ?

Relationships are the key to functional resilience

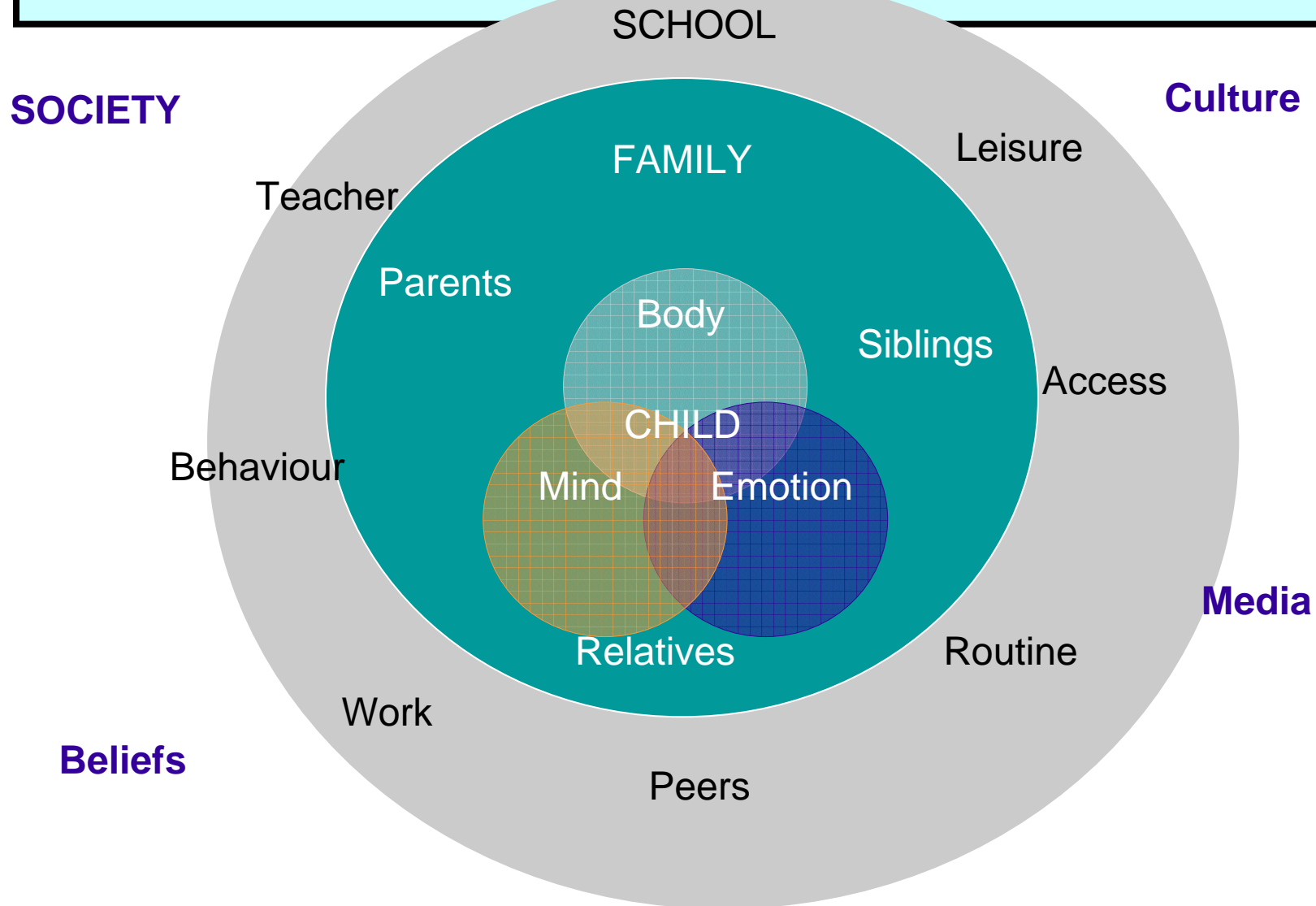
Established relationships

- * GP, paediatrician, other specialists
- * Local therapists
- * School, community supports

New relationships needed to accept help

- * Genetic
- * Medical
- * Emotional
- * Practical

Relationships - A child's world



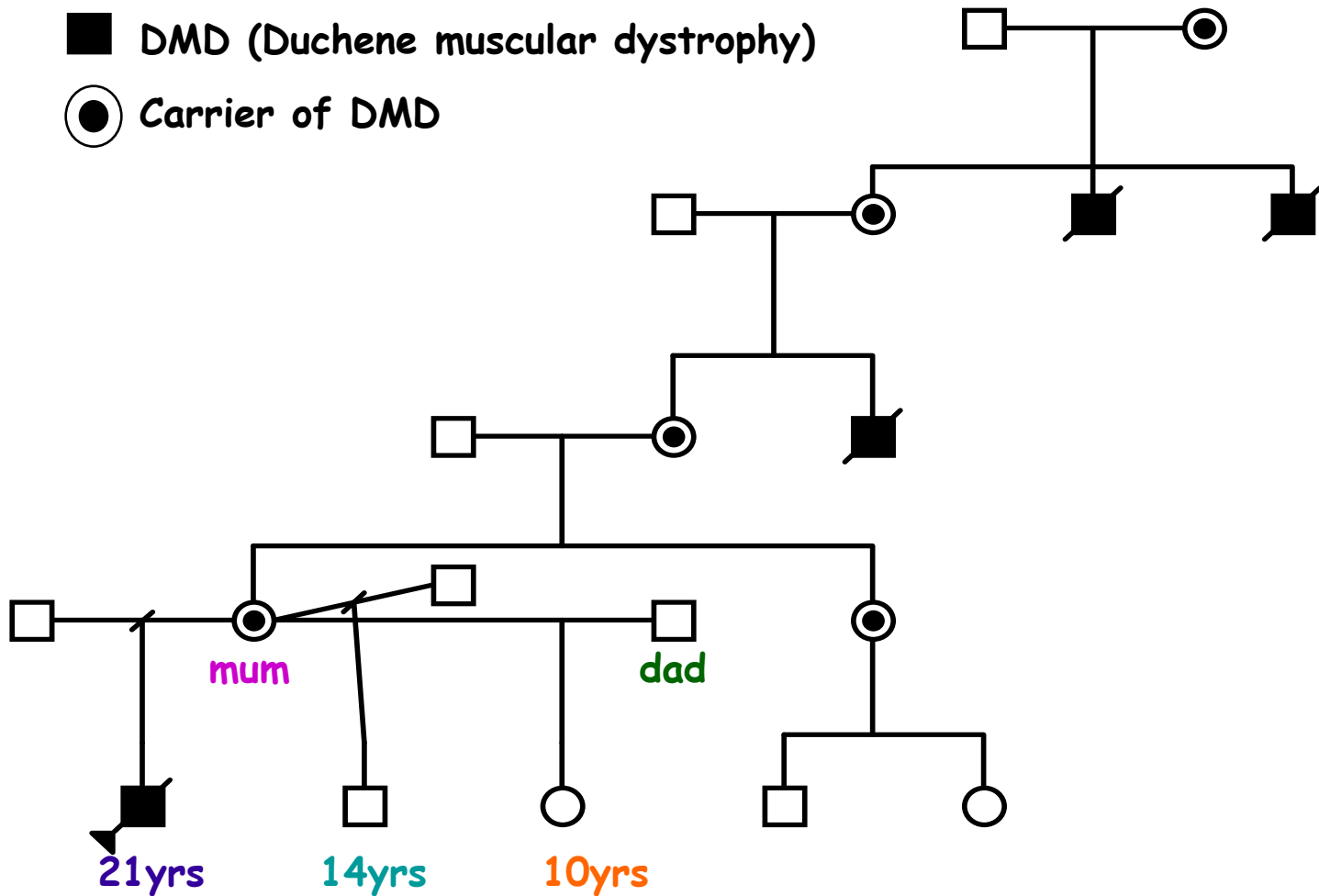
What 'help' do we offer families ?

- * Obvious help
- * Pre-emptive help
- * 'Helpful help'

Our family ...

■ DMD (Duchene muscular dystrophy)

○● Carrier of DMD



Our First contact with a family..

Intake: starts our relationship

- Provides initial information and support - family always very anxious about coming to genetics!
- Usually Genetic counsellor will make phone contact with family, often multiple times before appointment
- Gathers relevant information e.g. family history etc
- Provides strategies for anxiety management, support and issues of disclosure
- Describes process of diagnosis and management
- Describes team members and their roles

Obvious help - Genetic / Medical

genetic tests

family history

medical history

clinical examination

Results of investigations

summarise
why we
gather clues -
**demystify the
process**



DX

Obvious help - Genetic / Medical

- other family members - diagnosis, carrier
- prognosis - education
- treatment / clinical trials / management
- 'tick box' on paperwork
- With or without specific diagnosis - information and ongoing support to all family members
- reproductive planning options
- Helps facilitate other immediate needs e.g. school visit



Obvious help - emotional

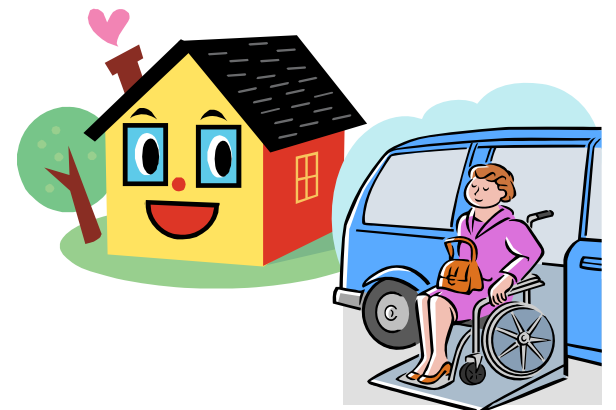


- counselling support for all family - grief, anger, fear
- decision making
- disclosure
- behaviour management
- mental health - sense of self
- motivation - 'why bother?'
- peer support / support groups / social support
- no fault for mutation or inheritance, guilt

Obvious help - practical



- therapy - OT, physio, social work, rehab
- accessing tertiary services - eg multidisciplinary clinics
- respite care / hospice / holidays / home care
- new systems - govt, funding organisations etc
- education - incl condition specific (centre for genetic education)
- travel
- financial
- dignity (avoid 'begging')



'Pre-emptive help..'

- overwhelming grief of Dx of condition - eg death sentence
- reproductive planning
- equipment and services
- management eg medicines (eg steroids), surgery, sleep therapy, clinical trials, research (international, multicentre)
- school / workplace / involvement
- 'make a wish'
- adolescent issues
- transition
- end of life issues



'Helpful help..?'

- evolving as family needs change
- enables compromise with dignity
- empowers
- averts :
 - Altered perception of genetic identity
 - Diminished perception of global health
 - Diminished social identity
 - Threat to parental role
 - Threat to family relationships



Self Concept - construct made up of cognitive, behavioural and affective components

Always back to ...

Why do we help..?



EMPOWERMENT

Our family..

Chronic sorrow coping strategies - emotional, interpersonal, cognitive, action.

Maintained their family relationships

Accepted their loss as a part of their new life

Survived the 'systems'

Maintained "hope"

**RESILIENCE
PREVAILS**