

Psychological impact of predictive genetic testing for arrhythmia disorders on children found to be predisposed to the condition and their families

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AIMS

- Arrhythmia Disorders
- Predictive testing (PST)
- Practical implications of PST
- Psychological impact of PST on:
 - Children
 - Parents/family
- Coping Strategies
- Implications for counselling

Arrhythmia Disorders

- Long QT Syndrome (LQTS)
 - Prevalence: ~1:3,000
 - Many different subtypes
 - Usually Autosomal Dominant inheritance
 - Incomplete penetrance
 - Variable expression
 - Genetic testing may confirm subtype – which can be useful for clinical management

Arrhythmia Disorders cont...

- Brugada Syndrome (BS)
 - Prevalence: ~1:2,000
 - Autosomal Dominant inheritance
 - Incomplete penetrance
 - Variable expression
 - Genetic testing will have little effect on clinical management but may enable cascade screening

Predictive Genetic Testing

- Possible when a causative mutation has already been identified in the family
- Ethical issues around PST in children
- PST in children is generally considered acceptable when there is possible onset in childhood/adolescence and when treatment or prevention is possible

Practical Implications

- Medical follow up and treatment
- Insurance
- Exercise
- Career
- Lifestyle

Psychological Impact: Children

- Can depend on background and timing
- Concern over possible serious consequences
- Concern for the future
- Feeling different from siblings/peers
- Wish to protect parents

Coping strategies (children)

- **Problem orientated**
 - Medication adherence
 - Limitation of physical exercise
- **Emotion-orientated**
 - Tried to stress similarities to peers, OR
 - Emphasise being different = special

Psychological Impact: Parents/family

- Mixed emotions
- Concern over possible serious consequences
- A balancing act
- Possible difference in levels of distress between parents and 'sides' of families
- How to manage the impact on family life when one child is a 'carrier' and another is not

Coping Strategies (parents/family)

- **Problem orientated**
 - Try to ensure medication adherence
 - Try to implement risk reducing behaviour
 - Put practical solutions in place
- **Emotion orientated**
 - Try to downplay family history
 - Try to implement risk reducing behaviour
 - Become active in patient support groups

Implications for counselling

- Ensure children have an accurate understanding
- Ensure children have a realistic notion of the controllability of their condition
- Have good communication with parents so you are aware of and can inform and support the choices they make

References

- Meulenkamp et al (2008) Predictive genetic testing for cardiovascular diseases: Impact on carrier children. *American Journal of Medical Genetics Part A* 146A: 31136-3146
- Hendriks et al (2005) High Distress in parents whose children undergo predictive testing for Long QT Syndrome. *Community Genetics* 8:103-113
- Smets et al (2008) Health related quality of life of children with a positive carrier status for inherited cardiovascular diseases. *American Journal of Medical Genetics Part A* 146A: 700-707
- Farnsworth et al (2006) When I go in to wake them... I wonder: Parental perceptions about congenital Long QT Syndrome. *Journal of the American Academy of Nurse practitioners* 18: 284-290
- Andersen et al (2008) Living with Long QT Syndrome: A qualitative study of coping with increased risk of sudden cardiac death. *Journal of Genetic Counselling* 17: 489-498
- www.geneclinics.org