Prevention of Thalassaemia: A challenge for improving patients quality of life

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Thalassaemia International Federation

“Equal access to quality healthcare for every patient with thalassaemia across the world”
Why prevention?

- Relevance to thalassaemia patients
- Relevance to prospective parents: at-risk couples
- Relevance to the community: public health issues.
Relevance to patients

- Optimum treatment is required for survival and quality of life.
- No treatment means early death.
- Less treatment means poor quality of life and early death.
Survival in thalassaemia

Modell et al. 1984

[Graph showing survival rates for thalassaemia patients with different treatments, comparing transfusion only, transfusion + Desferal, and no treatment.]
Survival of thalassaemia patients in Cyprus by birth cohort

Survival Functions

Birth Cohort

- 1980-
- 1980- censored
- 1975-9
- 1975-9 censored
- 1970-5
- 1970-5 censored
- 1965-9
- 1965-9 censored
Patient care

- Voluntary blood donation and adequate supplies
- Safe blood
- Provision of essential drugs chelating agents
- Free medical treatment
- Multidisciplinary care
Why prevention?

- High frequency of the condition in some populations, e.g. Cyprus 1:7 carriers

- Need to limit new cases so that resources can be available for patients

- The choice of the majority of at-risk couples. (Angastiniotis M et al. World Health Forum 1986)

Accumulation of cases

Assuming full survival and no prevention
Increase in blood requirements

If no prevention

Blood requirements units/year

0 10000 20000 30000 40000 50000
1 4 7 10 13 16 19 22

Blood requirements units/year
WHO estimated in 1971 that in the absence of effective prevention in Cyprus by 2010...

- Increased survival could lead to a rise in prevalence from 1:1000 to 1:138, which could result in an increase of 600–700%

More than 50% of the population would have to become blood donors
# Extent of Clinical Management and Prevention Across the World

<table>
<thead>
<tr>
<th>Country</th>
<th>Blood Transfusion</th>
<th>Iron Chelation</th>
<th>Prevention</th>
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<tbody>
<tr>
<td>Australia</td>
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<td>China</td>
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<td>Sri Lanka</td>
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<td>India</td>
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*Blood Transfusion* - National programmes of prenatal diagnosis and screening.

*Iron Chelation* - National programmes of prenatal diagnosis and screening.

*Prevention* - National programmes of prenatal diagnosis and screening.
Age Distribution of Thalassaemics in a country with no programme

Age Distribution Cyprus 1999
<table>
<thead>
<tr>
<th>Not surviving</th>
<th>Very cheap</th>
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<tbody>
<tr>
<td>Surviving with minimal treatment</td>
<td>Waste: dependent, invalid, early death</td>
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<tr>
<td>Surviving with complications</td>
<td>Medical costs much higher. QoL ↓</td>
</tr>
<tr>
<td>Surviving with optimal treatment and adherence to treatment</td>
<td>Contribution to economy, QoL ↑</td>
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What is Quality of Life?

- General well-being – happiness (subjective qol)
- Fulfilling goals and hopes (objective qol)
- Diverse and objective and subjective indicators
- Health related QoL: how QoL is affected by chronic illness.
- Achieving personal goals, hopes and aspirations – the happiness requirements
- The degree to which a person enjoys the important possibilities of life

www.utoronto.ca/qol.
How satisfied are you with your health

<table>
<thead>
<tr>
<th>Satisfaction Level</th>
<th>Percentage</th>
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<tbody>
<tr>
<td>Very dissatisfied</td>
<td>6.2%</td>
</tr>
<tr>
<td>Dissatisfied</td>
<td>11.3%</td>
</tr>
<tr>
<td>Neither dissatisfied or satisfied</td>
<td>36.1%</td>
</tr>
<tr>
<td>Satisfied</td>
<td>35.1%</td>
</tr>
<tr>
<td>Very satisfied</td>
<td>11.3%</td>
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</table>
## How would you rate your QoL

**Cyprus 2008**

<table>
<thead>
<tr>
<th>Rating</th>
<th>Percentage</th>
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<tbody>
<tr>
<td>Very poor</td>
<td>2.1%</td>
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<tr>
<td>Poor</td>
<td>9.3%</td>
</tr>
<tr>
<td>Neither poor nor good</td>
<td>32%</td>
</tr>
<tr>
<td>Good</td>
<td>35%</td>
</tr>
<tr>
<td>Very good</td>
<td>21.6%</td>
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</table>
Analysing QoL: becoming

- **Practical becoming**, purposeful activities, personal goals, wishes, working, learning
- **Leisure becoming**, relaxation, stress reduction, games, walks, visits, vacations
- **Growth becoming**, improving health, status, knowledge, skills, coping with changes
• Physical belonging, Home, environment, workplace, community

• Social belonging, being accepted, being intimate, family, friends, having a special person (spouse etc)

• Community belonging, access to education, work, income, recreation, social activities, access to services

Analysing QoL: belonging
• **Physical Health**, nutrition, exercise, appearance.

• **Psychological health**, adjustment, cognitions, feelings, moods, freedom from worry and stress, self evaluation

• **Spiritual being**, values, beliefs, hope for the future

Analysing QoL: being
Relevance to at-risk couples

- The right to healthy children
- The right to information
- The right not to know
- The right to choose
Couples who carry a genetic disorder

Reproductive options:

- Remain childless
- Take a chance, risk of having an affected child
- Prenatal diagnosis
- Pre-implantation Genetic Diagnosis (PGD)
- Gamete donation (artificial insemination)
- Adoption
Relevance to families

- Without a prevention program:
  - The possibility of having one (or more) affected child
  - Fear - leading to termination of pregnancies, 3/4 of which are of healthy fetuses
  - Death of affected children
  - Psychosocial burden
  - Economic burden
Public health

Cost-effectiveness of prevention:

- Cost of one year’s prevention = 8 week’s treatment (Angastiniotis M et al 1986)
- Lifetime healthcare cost of caring for one patient versus the cost of a national prevention program = 4.22:1 (Ginsberg G et al 1998)
Prevention strategies

- Public education and awareness
- Population screening. Specialised labs
- Genetic counselling. Counsellors?
- Prenatal diagnosis
- Pre-implantation diagnosis
- Ethical principles: voluntary, autonomy of couples, right to full information, confidentiality: informed choice
Health education / pre-screening counselling

- Education of the public – they must be aware of why they are being screened
- They must be told what thalassaemia is
- What is the life of a homozygote like
- What are the chances of being a carrier
- The consequences when two carriers marry
- The right not to know is respected

European Council Convention on Human Rights and Biomedicine
Thalassaemia genetic counselling

- Discuss the results of screening
- To know and understand their risk
- To assist them to make informed choices without being directive
- Discussion of options in a non-directive manner
- Respecting the autonomy of the couple
- This is the job of a specialist who knows thalassaemia
Genetic counselling session

When in a genetic counselling session, establish principles of:

- Privacy
- Trust
- Confidentiality
- Informed free consent
Residual Births Cyprus 1985-94
15 cases.
Conclusion

- Prevention benefits the patients by saving resources for their treatment.
- Early (before marriage) identification of carriers gives young couples the most choices.
- The health services can offer quality care so patients have quality of life as well as survival.
Prevention – is it Eugenics?

Concern because screening involves whole or parts of a population

Eugenics: “the improvement of the inborn qualities or stock of the human population”
(Francis Galton – Am J Sociol 1904, 10(1))

Aims at betterment through selection of ‘good genes’. Not eliminating disease but enhancing ‘normal’ traits

Coercion through laws and regulations
( Genomics and World Health – WHO, Geneva 2002)
Medical genetics vs. Eugenics

- Medical genetics aims to improve the lives of individuals and families
- It does not recognise ‘good’ genes and ‘bad’ genes
- Medical genetics recognises that diversity contributes to survival and the richness of humanity
- It does not use coercion