ETHICAL ISSUES IN GENETIC SCREENING FOR THALASSEMIA IN INDONESIA

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Outline

1. Introduction
2. Methodology
3. Results
4. Discussion
5. Conclusion

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Introduction

Evidence of Thalassemia
- The most common inherited single-gene disorder
- New cases: 2,500/ year in Indonesia

Treatments for Thalassemia
- Life-long dependence on blood transfusion and chelation therapy ($20-30 thousands/ patient/ year in Indonesia)

Prevention of Thalassemia
- Screening test, Genetic counselling, Prenatal diagnosis
- Involves issues of being inheritance, sharing genetic information, reproductive decision

Risks for Ethical Problems,
Essential for conducting an investigation

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Purpose of the study

Investigate the ethical issues raised by genetic screening for thalassemia by exploring the psychological and social implication of carrier testing and genetic counselling practices on people identified as carriers.
Methodology

Design
- A Phenomenology qualitative approach
- An exploration of individual lived experiences & seeking the meaning embedded

Sample
- Purposive sampling technique
  (Having children with thalassemia, Carriers, Having experiences in genetic screening & genetic counselling)
- Sample Recruitment: in the paediatric ward
- 6 parents (12 people) were invited
- 6 Mothers selected as participants

Data Collection
An in-depth interview with semi-structured questions taking roughly one hour

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Methodology cont…

Data Analysis
Braun & Clarke (2006) thematic analysis:

1. Familiarizing yourself with your data
2. Generating initial codes
3. Searching for themes
4. Reviewing themes
5. Defining and naming themes
6. Producing the report
Methodology cont...

Methodological Rigour
- Credibility
- Confirmability
- Transferability
- Fittingness

Ethical Consideration
- Respect for Persons (Voluntary participation, Informed consent)
- Beneficience (Favourable risk/benefit ratio)
- Justice (Fair selection, sharing of risk & benefit)
Results

1. Psychosocial reaction; between joy and sorrow

2. Negative social stigma

3. Fear of discrimination

4. Reproductive decisions dilemma

5. Truthfulness and braveness to share genetic information with the children

6. Religious belief influence regarding abortion decision in prenatal screening

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Theme 1
Psychosocial reaction; between joy and sorrow

- Participants expressed psychological distress, sadness, feelings of shock & fatigue

- Some mothers described their experiences as being positive

Understand more about thalassemia, being closer to God, more appreciative of the “little things in life”
Theme 2

Negative social stigma

Most of participants suffered from a social stigma because of being inheritance of the disease (thalassemia)

The stereotype of “the inheritance of thalassemia” revealed a meaning that “being a carrier of a chronic illness trait is a negative thing”
Theme 3
Fear of discrimination

- Participants expressed feeling “differed from others”

- Worried about social acceptance (if people would treat them differ from “normal people”)

- Some mothers worried about sustainability of social support and national policy
Theme 4
Reproductive decisions dilemma

❖ All participants stated they did not know that they or their husband were carriers of the thalassemia gene until they have children diagnosed thalassemia and got screening test for thalassemia

❖ Some respondents experienced dilemma for taking a decision to have another child (a dilemma between “happiness to have another child and fear to face burdens of care because of having children with thalassemia”)

❖ However, Some mothers believed that a child is a blessing from God, so they do not have to be worried for having another child
Theme 5

Truthfulness and braveness to share genetic information with the children

- Participants experience psychological challenges once they have to share genetic information with the children.

- It was hard for some mothers to tell the truth and be brave to explain that they were carriers of a chronic disease and they children would have risks to be a carrier or suffered from thalassemia.
Theme 6
Religious belief influence regarding abortion decision in prenatal screening

- Participants noted that the genetic counseling have informed them well about prenatal screening test.

- Some mothers explained their view that they may reject the termination of the pregnancy to prevent thalassemia for religious and cultural belief reasons.
Discussion

The main issues embedded in the themes are highlighted then the similar notions are gathered into a specific topic discussion.

Compared to previous study:
Prevention of thalassemia program is not widely applied in Indonesia

Similar to previous studies:
Screening test and genetic counseling have increased participants knowledge

Comprehensive understanding gained includes thalassemia disease, how to prevent thalassemia, benefits and procedure of screening test/ genetic counseling, how to gain access and share genetic information, and possible consequences being carriers.
Discussion cont...

Similar to previous studies:
Parents getting genetic screening identified as carriers experienced various psychosocial strains

In this study, positive impact is found

Society support is essential for releasing mothers’ strain

The process of identifying and labelling difference in the stigmatisation process is influenced by the social assumption about the particular chronic illness in the community
Spiritual beliefs is identified as the strength and core spirit

Abortion especially for reducing the birth of new cases of thalassemia is a sensitive issue widely debated in Indonesian society

Ethnicity and spiritual belief may influence mothers in the process of decision making (Reproductive decision and pregnancy termination decision if prenatal screening is conducted)
Conclusion

Screening test and genetic counseling could provide profound benefits.

Educating society is valuable for creating positive perception/assumption and building social support.

Prevention of thalassemia has psychosocial implication which may raise some ethical problems.

Screening test and genetic counseling applied should concern the Spiritual and cultural values.
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Thank You!