

Congenital and genetic disorders

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Description of condition and intervention

Congenital disorders may lead to infant and childhood mortality, lifelong illness and disability. Every year about 295000 newborns within 28 days of birth globally die due to congenital anomalies. These disorders not only have an influence on the affected individual, but also affect their families, health systems and societies as a whole. Genes have been implicated to play an important role in genetic disorders. Early detection of these disorders could be done at preconception, during pregnancy and in the neonatal period (WHO 2021).

Intervention attributes

Type of interventions

Prevention

Delivery platform

First-level hospital

Equity

In addition to considerations like cost-effectiveness and health systems factors, dimensions of equity can be relevant for priority setting. The opportunity for a long and healthy life varies according to the severity of a health condition that individuals might have, so there are inequities in individuals' opportunities for long and healthy lives based on the health conditions they face. Metrics used to estimate the severity of illness at an individual level can be used to help prioritize those with less opportunity for lifetime health. FairChoices: DCP Analytics Tool uses Health adjusted age of death (HAAD), which is a metric that estimates the

number of years lived from birth to death, discounting years lived with disability. A high HAAD thus represents a disease less severe in terms of lifetime health loss, while a low HAAD represents a disease that is severe on average, causing early death or a long period of severe disability. It is also possible to estimate the distribution of HAAD across individuals with a health condition. FairChoices shows for each intervention an average HAAD value of the conditions that are affected by respective interventions that have health effects. Additionally, a plot shows HAAD values for around 290 conditions (Johansson KA et al 2020).

Time dependence

Moderate level of urgency. Treatment outcomes not highly affected by some days of delay.

Population in need of interventions

Treated population comprises of prevalent cases of Thalassemias trait in the age group 15 to 49 years. The treated fraction is 1 for this intervention. We estimate only costs for diagnostic intervention.

Model assumptions

Table 1: Summary of model parameters and values used in FairChoices – DCP Analytical Tool

Category	Model parameter	Notes
Interventions	Congenital and genetic disorders: Identification of specific single-gene disorders and counselling	
Cost calculation		
Treated population	Prevalent cases	Global Burden of Disease Study 2019
Gender	Both	
Age	15 to 49 years	
Treated fraction	1	

Intervention cost

The cost of retrospective identification of carriers and prospective premarital screening and counseling to reduce conception rates, in settings where specific single-gene disorders are a public health concern, is estimated to be 100 USD per person-year in Iran's specified population in 2006.

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References

WHO 2021: Available from <https://www.who.int/news-room/fact-sheets/detail/congenital-anomalies>

Johansson KA et al 2020: Johansson KA, Coates MM, Økland JM, Tsuchiya A, Bukhman G, Norheim OF, Haaland Ø. Health by disease categories. Distributional Cost-Effectiveness Analysis: Quantifying Health Equity Impacts and Trade-Offs. 2020 Sep 30:105.

Ahmadnezhad E, Sepehrvand N, Jahani FF, Hatami S, Kargar C, Mirmohammadkhani M, Bazargan-Hejazi S. Evaluation and cost analysis of national health policy of thalassaemia screening in west-azerbaijan province of iran. Int J Prev Med. 2012 Oct;3(10):687-92. PMID: 23112894; PMCID: PMC3482995.

Appendix

Literature Review for effectiveness & safety

This literature search is an example of Level 1 search for intervention inputs taken from DCP3 or generated in an ad hoc manner (e.g., quick google search found one study of cervical cancer screening cost-effectiveness that was used to create an effectiveness parameter for that intervention).