Review of: "The Plight of Rare Diseases in Southern Africa: Health and Social Services Policy Recommendations"

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This article is reviewing rare diseases in the southern part of Subsaharan Africa-SSA- (SADC) with recommendations for policy makers. The recommendations include adopting a common definition, centralizing healthcare services, adopting preventive approaches, fostering collaborative research, and capacity building for healthcare workers.

For the definition, I recommend to separate monogenic diseases from others. As an example of the paper, multiple sclerosis (MS) should go to the group of immune mediated inflammatory diseases rather than to the rare diseases group. Several drugs may change the life of MS patients and possible low cost providers do exist. As an example, within the Accord initiative, Pfizer has committed to provide its entire portfolio including patented medicines and vaccines available in the U.S. or EU on a not-for-profit basis to the governments of 45 lower-income countries most vulnerable to healthcare inequalities to support patients through public health systems. This is applicable to several SADC countries.

I do not find in this paper what is specific to SSA except what is already known for neglected tropical diseases concerning poverty and limited access to health care and drugs. The problem of consanguinity in SSA is not reviewed, and would be worth of it for prevention and for targeting the policy, because recessive inherited diseases are in general more severe. For rare diseases black skin is already an important specificity to limit recognition of some birthmarks such as café au lait spots for neurofibromatosis. Also loss of pigment as in albinism leads to skin cancer in a far higher proportion as compared to countries of northern latitudes, and easier access to photoprotection and facilities for cutaneous surgery are needed.

Prevention for genetic disease needs already targeting a disease or a disease group. The case of albinism is paradigmatic because the diagnosis is obvious on black skin and that the disease is the cause of many societal problems around in SSA. Before offering a systematic screening, the possibilily of genetic counseling with a common molecular diagnostic platform for albinism in SADC could already be a significant initiative.

To conclude my recommendations, I think that for each medical specialty in SSA, the specificity and proritization of rare genetic diseases in the region should be first surveyed and analysed to make proposals for a common policy. Having a too broad scope will dilute the initiative, in the context of too limited resources for a broad coverage of rare diseases.