Sickle Cell Disease [1]

Novartis has long been committed to understanding and developing treatments for hemoglobinopathies, a group of inherited blood disorders, which includes sickle cell disease (SCD). SCD is a hereditary and life-threatening condition that causes chronic debilitating symptoms, including acute painful episodes, anemia, organ damage, chronic pain, and fatigue. It is regarded by the World Health Organization (WHO) as a public health priority and a neglected health problem in sub-Saharan Africa, where 80% of babies with SCD globally are born every year[1].

Today, Novartis is a leader in addressing critical unmet needs in SCD. We are continually seeking new approaches and strategic partnerships to help make diagnosis and treatment more accessible and affordable to patients in countries where SCD remains a challenge for health care systems.

Building a solid, sustainable foundation

SCD is a global health problem, yet sub-Saharan Africa bears the highest burden of disease. To help bridge disease management and outcome disparities between Africa and the rest of the world, Novartis established the Novartis Africa Sickle Cell Disease program. The program is implemented through public-private partnerships with local governments, as well as collaborations with universities, patient groups and other organizations. A key element of our strategy is a holistic approach to disease management that encompasses screening, diagnosis, treatment, education, research, and advocacy.

Video of Sickle Cell Disease

The program was first launched in Ghana [2] in 2019 and is now expanding to Uganda, Kenya and Tanzania [3], with further plans to reach at least 10 countries by 2022.

Highlights of the program include:

- Working with partners to make diagnosis more available, accessible and affordable—currently, lack of access to newborn screening, and thus delayed diagnosis, is thought to be a primary driver of mortality for children in Africa with SCD (it is estimated that 50-90% of infants born in Africa with SCD will die before their fifth birthday)[2]
- Expanding access to hydroxyurea (a generic medicine that has been shown to improve health outcomes of individuals living with SCD), as well as developing and making available a child-friendly formulation
- Strengthening the health care system for the comprehensive management of SCD, including the safe administration of treatments and monitoring of patients
- Partnering with governments for sustainability and to support policy change, as well as

prioritizing SCD in their health care agendas

Download the sickle cell disease fact sheet [4]

Nana is a sickle cell patient from Ghana

Enabling access to next-generation therapies

Novartis is also working to bring innovative therapies to sickle cell patients in sub-Saharan Africa. We plan to conduct two clinical trials, starting in Ghana and Kenya, to evaluate the safety and efficacy of a next-generation biologic therapy[3].

To help ensure that medicines are adopted and used successfully, we are supporting local partners to strengthen health care system capabilities, for example, supporting programs for training of health care professionals and the establishment of clinical centers of excellence.

Leveraging technology with the goal of improving outcomes and helping to optimize safety

Our comprehensive approach to SCD disease management includes a portfolio of digital initiatives, including information, advocacy, and support-oriented websites tailored to country-

specific needs, as well as mobile solutions to better serve the global sickle cell community.

For example, as part of the Africa Sickle Cell Disease Program, Novartis supported the Sickle Cell Foundation of Ghana to develop and roll out a digital phone-based application (app) to facilitate collection and management of data in their newborn screening program, as well as a clinical management app to help ensure that hydroxyurea is administered safely and that patients gain the maximum benefit from the treatment.

Addressing understanding: Making education a priority

There's a world of myths and misconceptions about SCD. Many of them are harmful and form barriers to early diagnosis and treatment of the disease. Novartis focuses on helping to educate patients and healthcare providers about the pathophysiology and management of SCD to confront barriers to adequate care and support. We work with partners to develop educational content and tools around our clinical trials for health care providers, who can share this information with patients.

Novartis has reached out to patients with the Sickle Cell World Assessment Survey (SWAY) [5] , one of the largest global surveys in sickle cell disease (SCD), which provided valuable information about the impact of the disease on patients and their families and caregivers worldwide[4]. Insights were collected from more than 2100 patients and 360 health care providers to help inform the management of this lifelong condition. Here are five things we learned [6].

Video of 5 things we learned about sickle cell disease from SWAY Survey

Additionally, we have long sponsored educational initiatives to bridge the voices of those with SCD with the world at large. One is example is notaloneinsicklecell.com [7], a website available in 15 different languages featuring real stories from patients with SCD from around the globe. We also use insights gathered from our patient advisory boards to create educational materials to support sickle cell communities directly. Finally, we collaborate with patient advocacy groups, in an ongoing capacity, to understand their educational goals and how they can best be met.

Gene therapy: Hope for a cure

In 2021, Novartis entered into a grant agreement with the Bill & Melinda Gates Foundation [8] with the shared goal to discover and develop a novel in vivo gene therapy to cure SCD.

Existing clinical-stage gene-based therapies involve removing cells from a patient, modifying those cells in a laboratory, and then reintroducing them to the patient through a procedure similar to a bone marrow transplant. If approved, this complex procedure may be challenging to introduce in many parts of sub-Saharan Africa due to current limitations of local healthcare systems. The Novartis-Gates Foundation collaboration envisions a single-shot in vivo gene therapy administered directly to patients, where the goal would be to mitigate the need for extended hospital stays or specialized infrastructures.

As part of the early drug design strategy, Novartis will prioritize addressing access and distribution hurdles posed by limited healthcare infrastructure in low- and middle-income

countries. The funding agreement includes specific provisions to support global access to resulting innovations.

At Novartis, we remain committed to providing hope and improving outcomes for anyone affected by SCD. There is more to be done. That is why we continue to join forces with partners from around the world to help bring sickle cell disease out of the shadows.

	**Partners** **Novartis partner, Hemex Health, develops point of care diagnostic** Watch Video [9]

News

- Novartis expands Africa SCD program to East Africa [3]
- Government of Ghana makes hydroxyurea available to people with sickle cell disease through the first of its kind public-private partnership with global medicines company Novartis [2]
- Improving the lives of people with sickle cell disease in Ghana [10]
- Government of Ghana launches first of its kind public-private partnership with Novartis to improve diagnosis and treatment of people with sickle cell disease [11]

Footnotes:

References:

1. Piel FB, Patil AP, Howes RE, Nyangiri OA, Gething PW, Williams TN, Weatherall DJ, Hay SI. Nat Commun. 2010 Nov 2; 1:104.
2. Grosse SD, Odame I, Atrash HK, et al. Sickle cell disease in Africa: a neglected cause of early childhood mortality. Am J Prev Med 2011; 41:S398.
3. www.clinicaltrials.gov [12], accesses last june 2021
4. Osunkwo et al. Am J Hematol 2021 96(4):404-417

Links
[1] https://www.novartis.com/our-impact/global-health/sickle-cell-disease
[2] https://www.3blmedia.com/News/Government-Ghana-Makes-Hydroxyurea-Available-People-Sickle-Cell-Disease-Through-First-Its-Kind

[3] https://www.novartis.com/news/media-releases/novartis-expands-africa-sickle-cell-disease-program-uganda-and-tanzania
[4] https://www.novartis.com/news/media-library/reimagining-management-sickle-cell-disease-africa
[5] https://www.novartis.com/news/media-releases/novartis-global-survey-uncovers-profound-and-often-under-reported-effects-sickle-cell-disease-patients
[6] https://www.youtube.com/watch?v=kJNCIlPeTeg
[7] https://www.notaloneinsicklecell.com/
[8] https://www.novartis.com/news/media-releases/novartis-and-bill-melinda-gates-foundation-collaborate-discover-and-develop-accessible-vivo-gene-therapy-sickle-cell-disease
[9] https://www.youtube.com/watch?v=BSUU2J6uQ4M
[10] https://devex.shorthandstories.com/sickle-cell-disease-in-Ghana/index.html
[11] https://www.3blmedia.com/News/Government-Ghana-Launches-First-Its-Kind-Public-Private-Partnership-Novartis-Improve-Diagnosis
[12] https://www.clinicaltrials.gov