spotlight

PABLO ALBARENGA FOR *NATURE*

Aline Saliba loves understanding the science behind paediatric medicine.

HOW PAEDIATRICIANS ARE ADVANCING CHILD HEALTH

Five clinicians describe how their research is contributing to a United Nations goal to boost health and well-being. **By Nikki Forrester**

One of the Sustainable Development Goals set by the United Nations is to ensure healthy lives and promote well-being for all people, in part by lowering global maternal mortality and ending preventable deaths of newborn babies and children under five years old. Advances in paediatric care over the past several decades have led to marked declines in child mortality. Between 1990 and 2019, global under-five mortality decreased by 59%[1], but millions of children still become sick and die each year, often from preventable and treatable causes (see 'Meeting the target'). These deaths occur disproportionately in sub-Saharan Africa and south Asia, which together accounted for more than 80% of the 5.2 million deaths of children under five years old in 2019, despite the regions comprising only 51% of the global population for this age group[1].

Paediatricians have a crucial role not only in caring for sick children and their families, but also in monitoring a child's development from birth to adulthood. They often conduct research to uncover the mechanisms that drive disease, develop therapeutics and tackle societal and environmental challenges that prevent children from reaching optimum health. *Nature* spoke to five paediatricians about what inspired them to pursue the speciality, their research programmes and how they are helping to improve child health around the world.

spotlight

ALINE SALIBA
PROVIDING CARE UNDER PRESSURE

After my six-year medical-school programme at the Catholic University of Brasília, I did two residencies at local hospitals in paediatrics and paediatric intensive care, and fell in love with how resilient and strong children are. Paediatric intensive care has a really low death rate in Brazil, around 5% (ref. 2). When I go to work, I know that I'm going to solve 95% of the cases that day. For instance, sometimes I see children with severe disease, and in a couple of days, they're smiling. I get a little emotional about it; it's magical to see those transformations.

I usually work shifts of 12–24 hours. Some days, everything is under control, but most days I see stressful events. I'm trained to recognize severe disease, such as respiratory distress or heart failure. But what makes parents most nervous is fever.

I really like how dynamic my job is. Every day is a surprise. I get to the hospital and I have no idea what the day is going to be like. Paediatrics is one of the most complex specialities in medicine because it encompasses a lot of different populations. It includes very premature babies, newborns, toddlers, children and teenagers. I see so many different people with different pathologies, diseases and physiologies.

One of the most challenging aspects of my job is dealing with the families of very sick children. Even if I know that things are probably going to be okay, I'm dealing with people who are really fragile and scared. Parents aren't used to being in the intensive care unit, which is a terrible place to be when you're not sick. It's cold, bright and noisy, and everyone is working so quickly. Sometimes it's hard for me to express to parents that I also feel scared and insecure. I don't always have time to embrace and comfort children and their families.

When I finished my residencies in 2015, I thought now I can rest. But in 2016, I decided to pursue a master's degree in congenital heart disease at the University of Brasília. I finished my master's in 2019 and began my PhD in 2020, studying how genetics influences congenital heart disease and how children respond to the surgical procedures for treating it. I've always loved understanding the science behind the medicine. Doing so allows me to view my work in the intensive care unit in a different way. It helps me to understand why a heart is failing and why a medicine isn't working.

Aline Saliba is a critical-care paediatrician at the Children's Hospital of Brasília José Alencar in Brasília, Brazil.

HANNA-KATRINA JĘDROSZ FOR *NATURE*

Ameenat Lola Solebo feels that child health is often an afterthought for public health.

AMEENAT LOLA SOLEBO
PUTTING CHILDREN FIRST

I support the care of children with glaucoma, cataracts and chronic inflammatory eye disorders that put them at risk of blindness. I spend most of my time working on a disorder called uveitis. Uveitis is a chronic inflammatory disorder of the eye that potentially causes blindness. It affects around one in every 1,000 children[3]. Some of the most effective therapies, such as steroids, come with a bucketload of side effects, so I have to be judicious when prescribing them to stop children from losing their vision. We definitely need a better understanding of the mechanisms of eye disease and how to target treatments.

During my training at Great Ormond Street Hospital in London, it was phenomenal to watch children grow and develop, and to be the clinician for part of their journey. I also love how families interact with clinicians. They're invested in that relationship, and then, as the child gets older, they join that relationship.

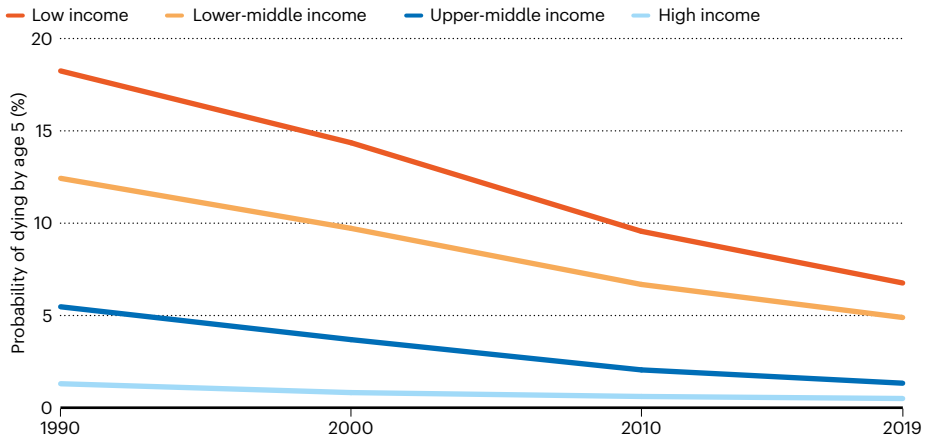
I spend about 30% of my time in clinical practice and the rest doing research. One of the things I'm really proud of is doing innovative paediatric science. Normally, you study a disease in adults and then see how it applies to children. But my colleagues and I are doing imaging work to study uveitis in kids first, despite the fact that the disease affects children and adults.

> **"I also love how families interact with clinicians. They're invested in that relationship."**

We're also running a national cohort study at 33 hospitals across the United Kingdom to try to understand how we can predict who is severely affected by uveitis and who isn't. A good outcome is when the disease is picked up before it causes problems with eyesight, or when a child responds to treatment quickly and the disease goes into remission. A poor outcome is when a child has limited vision, either because of delays in diagnosis or because they didn't respond well to treatment.

MEETING THE TARGET

The risk of death in children under five fell steadily between 1990 and 2019 across 195 countries. Millions of children still die every year, however, particularly in low- and lower-middle-income countries. If the United Nations goal to reduce under-five mortality is met, 11 million deaths could be averted between 2020 and 2030.



SOURCE: REF. 1

One of our early findings was that general, family and primary-health-care practitioners need greater support to recognize signs of uveitis early on[4]. It's an uncommon disorder in children, so practitioners might only see one case every few years. My colleagues and I are developing resources to help them to identify these rare diseases, including writing an article on how to look for changes in the eye.

Unfortunately, child health seems to be an afterthought when it comes to public health and clinical practice. Children are not mini adults. Disorders present differently in children. So much of the work that's being done in ophthalmology focuses on later-life adult diseases, such as cataracts, glaucoma and age-related macular degeneration. Unless we have researchers focusing on what disorders look like in childhood and how they affect a child's development, quality of life, socialization and other outcomes that actually matter to children, then we're not giving them the best chance to be the best adults they can be.

Ameenat Lola Solebo is a paediatric ophthalmologist at Great Ormond Street Hospital for Children, London, and a research clinician scientist at University College London Great Ormond Street Institute of Child Health.

MAYA CHOPRA
DIGGING INTO RARE DISEASE

When I rotated through paediatric specialities during my training at the Children's Hospital at Westmead in Sydney about 20 years ago, I was drawn to the difficult, complex cases.

I saw individuals who had rare disorders and observed members of the genetics team when they gave their opinion. They would spend an hour with the patient, review the literature and come back with a report. The first time I read one of those reports, I knew that was what I wanted to do.

I was fascinated by the depth of literature they reviewed and evaluated in the reports. I liked the idea of putting pieces of a puzzle together and seeing how a pattern of clinical features led to a set of diagnoses. The field has moved so fast that we're now able to sequence whole genomes and combine those data with clinical information to form an opinion.

Rare diseases have an enormous public-health burden. For example, one in ten families in the United States is affected by a rare disorder[5], and 80% are genetic[6]. Sometimes a disease is so rare that there might be only a handful of other cases in the world. It can be very challenging and isolating for an individual and their family to find out that they're the only person in the state or in the country with that condition.

In my current role I work on advancing therapies from the research bench to the clinic for rare genetic disorders that affect neurodevelopment. The disorders can lead to conditions such as intellectual disability, autism spectrum disorder and epilepsy, which collectively affect 1% of the population.

I also run my own research programme at Boston's Children's Hospital, Massachusetts, studying a rare genetic disorder, called Chopra–Amiel–Gordon syndrome, that is characterized by intellectual disabilities, speech delay and particular facial features[7]. I co-discovered this disorder when I was working at the Imagine Institute of Genetic Diseases in Paris. In 2021, my colleagues and I authored the first study of an international cohort of 34 people with the disorder[8]. This meant that it could be registered as a disease and individuals could be diagnosed.

Along with interacting with families through my research programme, I spend a lot of time with patient advocacy groups for rare genetic disorders. These groups are leading the way in providing a patient voice when it comes to advocating for their disorder to be considered for therapeutic development. They also inform researchers about what the

BOSTON CHILDREN'S HOSPITAL

Maya Chopra says that patient advocacy groups are key to deciding treatment priorities.

priorities should be, because when we talk about treating a disorder, we need to think about what exactly we are trying to treat. Is the goal to extend life, improve communication or manage a behaviour such as sleep? As researchers, scientists and physicians, we need to hear about what is important to the people with a given disorder.

Throughout my career, I've worked in Australia, China, France and the United States. Despite the differences in cultures and health-care settings, families all have the same questions. They want to know what a diagnosis means for their child and what the pathway is to treatment. At our core, everybody wants access to the best knowledge and technology to be able to care for children.

Maya Chopra is director of the Translational Genomic Medicine Core of the Rosamund Stone Zander Translational Neuroscience Center and a clinical geneticist at Harvard Medical School in Boston, Massachusetts.

ORODE DOHERTY
EXPANDING ACCESS

As an infant growing up in Lagos, I was very sickly. I was at hospital all the time because I had febrile convulsions. I was inspired by the people I saw at the hospital. As I got older, I had the unofficial role of helping my family members to get to the health centre, around a 30-minute walk from our home, when they had an injury. Those experiences motivated me to pursue medicine at the University of Benin, Nigeria, in 1986, and then two paediatric residencies, in Nigeria and the United States.

Nigeria has one of the highest maternal and newborn death burdens in the world[1,9]. Many women avoid going to hospital or are delayed getting there, leading to avoidable deaths. Mothers might not have access to safe, skilled and trustworthy health providers. My work at Ingress Health Partners, a primary health-care company I founded in June 2020, includes training nurses and birth attendants to ensure that women receive safe services and that those services are escalated as quickly as possible when issues occur.

Another challenge with children's health in Nigeria is that the country is not very good with preventive care. All the basic immunizations are given, but otherwise, health-care providers typically see children only when they're sick or if they have a severe vulnerability, such as being born preterm. Ingress Health Partners promotes preventive care and anticipatory guidance for families.

> **"Mothers might not have access to safe, skilled and trustworthy health providers."**

This means that we educate parents on what to expect for every phase of a child's life. For instance, I teach mothers how to breastfeed and check newborn babies' eyes to make sure they're not yellow, which might need to be

ANDREW ESIEBO/PANOS PICTURES FOR *NATURE*

Orode Doherty started a company to provide people with safe maternity services.

treated to avoid brain damage. I also advise families on what behaviours to expect from their babies at 6 weeks, 10 weeks and 3 months, for instance. If there are any red flags, parents know to come back for a check-up.

In a country where poverty is rife, basic health-care access and financial constraints can mean that children don't get the care that they need. I'm working with other health-care organizations and policymakers to expand access to universal health coverage[10]. We have come up with a model that we've seen work elsewhere, and we're looking forward to piloting that.

Orode Doherty is founder and chief executive of Ingress Health Partners in Lagos, Nigeria.

ANDREW BECK
MERGING SOCIAL SCIENCE AND MEDICINE

NIKKI FORRESTER FOR *NATURE*

Andrew Beck's degree in anthropology has helped his research on health inequities.

My inspiration to go into paediatrics was driven by my experience as a counsellor at Camp Seneca Lake in upstate New York in the late 1990s. I love working with kids. Being part of an incredible experience at the summer camp for young people is not all that different from my role as a paediatrician — I try to help children to have the best chance of being healthy, thriving and reaching their full potential.

At medical school at the University of Pittsburgh in Pennsylvania, paediatric rotations were my favourite. In the clinic, about half of the kids I see now come in for a preventive visit or a check-up to have their immunizations, developmental screens or physical examinations to ensure that they're growing and developing as expected. The other half are usually there for a health complaint, such as a fever, a cold, asthma or pneumonia.

Much of my work as an attending physician and academic researcher focuses on the intersection of place and health. This is driven largely by the disparate environments in which children and their families are born, grow, live, work and age. These factors can include healthy housing, food security, social capital and exposure to support systems, all of which can be driven by structural racism and economic disinvestment.

One way we've tried to enhance environments is through the Cincinnati Child Health-Law Partnership. This is a programme that was developed about 15 years ago to bring clinicians together with legal advocates who are specialists in the civil and legal rights of low-income individuals and families. Every year, about 1,000 families from Cincinnati Children's primary-care centres are referred to this programme. The attorneys and paralegals help them with issues such as adverse housing conditions, public-benefit denials or delays, and unmet educational needs. Their advocacy has improved housing conditions and recovered benefits that resulted in hundreds, if not thousands, of dollars going back into the pockets of families.

"Referrals to the partnership programme had driven down hospitalization rates by nearly 38%."

Last year, we reported[11] that referrals to the partnership programme had driven down hospitalization rates by nearly 38%. Currently, we're exploring how to extend this partnership beyond primary-care centres to specialty clinics across the hospital.

My undergraduate degree in anthropology is incredibly relevant to my role as a paediatrician. In examination rooms, it helps me to contextualize the care that I provide. In my academic life, it helps me to think about what studies we can do to evaluate underlying inequities in health outcomes and potential interventions we can put into place to move towards better, more equitable outcomes for children in our community. Bringing a medical or public-health lens to questions around race, poverty or discrimination is really important because we won't be able to solve the medical challenges that are ever-present in the clinic without enhancing the environments in which the kids and their families live.

Andrew Beck is a paediatrician at Cincinnati Children's and a paediatric researcher at the University of Cincinnati College of Medicine in Ohio.

Interviews by Nikki Forrester.
These interviews have been edited for length and clarity.

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Correction

This Spotlight article erroneously referred to Maya Chopra as a neurologist. In fact, she's a geneticist. Also, Chopra is running her research programme at Boston Children's Hospital, not Harvard Medical School. Finally, the article misstated the extent of the advances made in genetic sequencing.

Corrected 8 November 2023