Fatma Al Jasmi – Biochemical Genetics Consultant; Dean, College of Medicine & Health Science, UAEU

The most pressing challenge, and one that transcends any single country, is the cost of advanced therapies

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Prof. Fatma Al Jasmi, Professor of Biochemical Genetics at the UAE University's College of Medicine & Health Science and the UAE's first biochemical genetics consultant, has spent nearly two decades building a rare and genetic disease infrastructure from the ground up. Prof. Al Jasmi traces that journey – from persuading hospital management, establishing national screening programmes, building a genomics research laboratory, and harnessing artificial intelligence for early diagnosis – and reflects candidly on what remains to be done.

You trained in Canada and returned to the UAE in 2009. Can you take us back to the beginning of that journey?

If you understand where I came from personally, you will understand everything that followed. I am actually a graduate of UAE University, the very institution where I now serve as dean. As a medical student here, what struck me immediately was the volume of genetic metabolic disease cases presenting in our hospitals, and the near-total absence of diagnosis or specialist expertise. My field ,biochemical genetics, the metabolic side of the discipline, simply did not exist here as a clinical speciality.

I became involved early on with a case that ultimately ended in the patient's death without a diagnosis ever being established. That experience shaped everything. When I left for Canada to pursue postgraduate training, I knew from the outset that I wanted to specialise in metabolic disease. I chose Toronto because of the presence of Dr. Joe Clarke, who had built the field there. It was an extraordinarily competitive environment; one fellowship position was available every three years, and I was fortunate to secure it. I completed my paediatric residency and then specialised in metabolic disease before returning to the UAE in 2009 as the country's first biochemical genetics consultant.

There was no one else in the country with that speciality. When I arrived, I was told that one clinic per month would be more than sufficient for such a rare field. Today, we have more than ten consultants running daily metabolic and genetics clinics, with long waiting times – a testament to the true burden of disease that was simply invisible before.

What did building that infrastructure from scratch actually involve?

Everything had to be created. I began at our teaching hospital, Tawam Hospital, establishing both inpatient and outpatient services, building the clinical team, developing the laboratory capacity to support diagnosis, and then working to ensure that new treatments were made available nationwide. I had to build global networks because in rare diseases, you have no choice. A highly specialised diagnostic test may be available at only one laboratory worldwide, such as in Australia or the US. I established those connections, ensuring diagnostic access and treatment availability for our patients.

But I realised early on that I could not do this alone, and that sustainable progress required a workforce. That brought me to my role at the university. I worked to establish the Department of Genetics and Genomics at UAE University, which did not previously exist, along with a Master's programme in genetic counselling and a Master's in genomic medicine. I worked with the teaching hospital and health authorities to establish Emirati fellowship programmes in clinical genetics, because the global shortage of clinical geneticists and genetic counsellors is acute, and we had to develop our own pipeline. We currently have around six fellows in training and have already graduated two Emirati consultants who are now serving in the field.

Simultaneously, I began working with the Ministry of Health on prevention, because for the majority of rare genetic diseases, no treatment exists. The burden on families and on the health system is enormous, and the most cost-effective investment is prevention. When I joined in 2009, newborn

screening was limited only to one metabolic disorder: Phenylketonuria (PKU). We expanded that biochemical screening to cover 40 metabolic diseases. More recently, the Abu Dhabi Newborn Genome Screening pilot program expanded to cover hundreds of actionable conditions. In parallel, the premarital screening program was scaled up from a pilot initiative to a national level, supported by the Emirati Genome Programme and incorporating whole-genome sequencing. This places the UAE among the leading countries globally implementing genome-based premarital screening at scale.

The premarital screening programme is now mandatory nationally. What were the challenges in getting there, and what cultural dynamics did you have to navigate?

It was one of the most significant projects I have worked on, and not without considerable difficulty. Implementing a national prevention programme requires preparing the frontline, in this case, family physicians who provide counselling and deliver results to couples. We designed training programmes at the UAE University covering the fundamentals of genetic disease and the practical skills of genetic counselling, with both theoretical and practical examinations. Only upon passing those assessments were physicians authorised to implement the programme. The rollout from piloting to national expansion took time, but it is now operating nationwide.

The cultural dimension was perhaps the more sensitive challenge. When I returned in 2009, there were people who said plainly that it would be impossible to change marriage practices in a region where consanguinity is deeply embedded in tribal culture and has been for hundreds of years. What I found, working directly with affected families, was that the scepticism dissolved the moment people were counselled properly and given real information. Families who had a child with a genetic disease came to understand that prevention was possible. And crucially, the government supported them: prenatal genetic testing and pre-implantation genetic diagnosis are all covered for families at high genetic risk. Those families pursued these options, had healthy children, and came back, not only accepting the programme but actively requesting genetic testing for their wider family. That shift in community engagement has been one of the most rewarding things I have witnessed.

In terms of early results, what are you seeing from these programmes so far, and how are you measuring their impact?

It is still early to draw definitive conclusions at the national scale, but the data we have is striking. In Western populations, studies of carrier rates for common genetic diseases in pre-conception screening range around two percent. Based on our pilot data, that figure reached 14 percent across the general population, and approached 25 percent in areas of high consanguinity.. That tells you precisely why a national programme in the UAE is not simply desirable but essential, and why it is more cost-effective per case prevented than in populations with a lower burden of inherited disease.

At the clinical level, what I can tell you is that recurrence within families I have been following families where a child with a genetic disease was previously born has declined markedly as those families have engaged with prevention pathways. That is a meaningful signal, even before the national programme has had time to generate population-level data.

From your experience, what stands out about the genetic makeup and disease patterns in the UAE?

The consanguinity and tribal structure of this region, sustained across hundreds of years, create a genetic disease profile that is genuinely different from anything I encountered in Canada. What I began to observe when we developed in-house diagnostic capacity was that patients here frequently present with not one but two genetic diseases simultaneously. In some cases, three. That produces a blended phenotype, a clinical picture that does not match the classical presentation of any single condition, which makes diagnosis extraordinarily difficult if you are working from a Western reference framework. The more we sequence, the more we see that co-occurrence of multiple genetic conditions within a single individual is far more common than the literature suggests. It fundamentally changes how these cases must be managed, and it underscores why research on this specific population is so important.

We have diseases present here in numbers that exceed global case counts for conditions that the wider world considers vanishingly rare. As a small Emirati population, we sometimes have more cases of a particular disease than the rest of the world combined. That is not a burden to be minimised – it is a scientific resource, and we are working to develop treatments for these ultra-rare conditions.

You have established in-house genomic sequencing and a national research study for unsolved cases. Can you tell us more about the research infrastructure you have built?

We now conduct whole exome and whole genome sequencing entirely in-house, both within the clinical service and through the UAE Genomics Laboratory that I established on a research basis at the university. The two functions are deliberately integrated because, in rare disease medicine, the boundary between clinical care and research is often illusory; some patients cannot be diagnosed through standard clinical testing, and further research is the only path to an answer.

To address this, we established a national research study that engages geneticists nationwide. When a case has exhausted all standard clinical and genetic testing and remains unsolved, it is recruited into the study for deeper in-house analysis. Our master's students in genomic medicine and genetic counselling are actively involved in this work as part of their training. It is a model that simultaneously builds the workforce and advances the science.

As a physician, clinician, and mother, I know that every family wants healthy children. That is what we are working towards. And there are two ways to achieve it: prevention, through national premarital and newborn screening, and early diagnosis, so that when disease does occur, we intervene before irreversible damage is done. Both pathways require continuous, dynamic research. Each population carries its own unique pathogenic variants, and ours is no exception. We hold regular meetings with the Department of Health to review and update the list of diseases covered by our screening programmes and to refine the specific mutations we are testing for. The more we test and learn, the better our outcomes become across prevention, diagnosis, and treatment.

One of the most important collaborations we have developed is with Khalifa University, where our team is working on artificial intelligence approaches to rare disease diagnosis using electronic medical records and generative AI to support earlier identification of these conditions by physicians. Several publications are imminent from this work. The focus is deliberately on treatable diseases, because the tragedy in rare disease medicine is not only the absence of treatments but the frequency with which patients are diagnosed too late to benefit from the treatments that do exist.

How do you approach collaboration with the pharmaceutical and diagnostics industry – a relationship that can be complicated in some settings?

I believe in it completely, and I always have. In rare disease medicine, no single institution, country, or sector can solve these problems in isolation. The diseases are too rare, the science too complex, and the resources required too great. Throughout my career, I have worked closely with the industry to improve diagnostic access and secure treatment for patients. In rare diseases, the global community is genuinely small; we know each other, we depend on each other, and we have to work across borders to give patients access to diagnosis, therapies, and clinical trials

The Department of Health and our health authorities have consistently supported compassionate access and facilitated participation in clinical trials when standard treatment is unavailable. I have patients who have accessed clinical trials abroad with transformative results. In one family, an older sibling was wheelchair-bound, while the younger sister is now mobile, developmentally on track, and indistinguishable from her peers, following early access to a study. That is what this work is ultimately for. Those are the stories that sustain you.

Your country is nearly 90% percent expatriate. How does this ecosystem serve the non-Emirati population?

With genuine equity, in my experience. When I returned in 2009, diagnostic access and treatment access were the same for Emirati and non-Emirati patients. That principle has not changed. The majority of my patients today are non-Emirati, and they receive the same diagnostic evaluation and the same access to treatment. The UAE’s model of healthcare provision for residents is, in my view, among the most equitable I have encountered anywhere in the world. There are now regulatory frameworks to ensure that access is appropriately prioritised for residents of the Emirates, and within those parameters, the commitment to serving everyone who lives here remains firm.

Looking forward, where are the gaps, and what still needs to be done?

To put it in perspective: although genomic sequencing has advanced dramatically, much of the genome’s functional meaning remains unknown. That puts into perspective how much remains to be discovered. The diagnostic odyssey – years of inconclusive testing before a diagnosis is reached – has shortened dramatically with the advent of whole genome sequencing, but it has not ended. Many cases remain unsolved. Many diseases have no approved treatment.

The most pressing challenge, and one that transcends any single country, is the cost of advanced therapies. As a clinician, I will always advocate for my patients to access the treatments they need. But the economic realities of gene therapies and other advanced interventions create genuine barriers, and finding ways to develop treatments that are effective, accessible, and affordable is a problem the entire global community of rare disease medicine must address together. Regulatory pathways for gene therapy approval must continue to evolve and accelerate. There is more research to be done, more treatments to be developed, and more international collaboration to be built. Our patients deserve a healthy life – and that aspiration does not stop at any national border.

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