JHARG: A Journal is Born!

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Born into a cold room

I want to begin with the truth, because an editorial that opens with pleasantries would be the first dishonest thing printed in this journal, and I would rather it were not.

JHARG has not had an easy birth. It did not come early. If anything it is overdue, carried far longer than it should have been, and being late has not made it any stronger. As Editor-in-Chief of the Journal of Health Advancements, Research and Genomics (JHARG), the official journal of the Society of Novel and Rare Diseases (SNARE), I am supposed to say that it is a privilege to welcome you to this maiden issue. It is a privilege. For most of the past many months it has also been a lonely one.

Support for this journal has been lukewarm at best. Meetings were held, approvals were given, good wishes were offered, and then very little happened. Emails went unanswered. Deadlines slipped and nobody seemed to mind. Somewhere along the way JHARG stopped being the society's project and became, in practice, mine: my worry, my late nights, my personal responsibility. I am not writing this to settle scores. Most of the people I have in mind are decent, overworked colleagues with crowded clinics and too many committees. I am writing it because it points to something bigger and sadder. In our hospitals and universities research sits at the very bottom of the pile. It comes after service, after administration, after the next inspection, after everything else. We praise it in speeches and starve it of time and money. A journal cannot live on speeches.

There is an image I cannot shake off. Before the Prophet Muhammad (peace be upon him) came, it was the custom among some Arab tribes to bury newborn daughters alive. The Quran keeps the memory of that cruelty in one haunting question, in which the buried girl is asked for what sin she was killed (Al-Quran, At-Takwir 81:8–9). I often think of the mothers in those households. A mother would hold her baby, feed her, love her with everything she had, all the while knowing what the family had already decided. She could dote on her child. She could not save her. That is uncomfortably close to how it has felt to bring JHARG into the world. You give yourself completely to something while suspecting that the people around it will let it die. Not out of malice. Out of indifference, which in the end does the same work.

And there is a further sadness. This child is not even growing up in its own home. Rare diseases are often called orphan diseases, and it is fitting, if painful, that a journal devoted to them should begin its life in a foster home, looked after by people who did not ask for it and are not yet sure they want it. Foster children learn early to keep quiet, to be grateful and not to ask for much. I do not want that for JHARG.

So we will raise it anyway, against the odds. JHARG is meant to become a meeting point for research, for the making of new knowledge, for the understanding of science, rare diseases and genomics, and for the advancement of healthcare. That breadth is deliberate.

Socrates,[1] whom the oracle at Delphi is said to have named the wisest of men, said, “True knowledge exists in knowing that we know nothing.” That humility, the willingness to doubt what we think we know and test it against evidence, is where all research begins.

Nowhere is our ignorance greater than in rare diseases. Each is uncommon, yet together they affect an estimated 263–446 million people; about 72% are genetic and 70% begin in childhood.[2] Pakistan carries more than its share, because consanguineous marriage raises the chance that a child inherits two copies of the same rare variant.[3,4] The same fact makes our population scientifically valuable. Sequencing of more than 10,000 Pakistani adults found “human knockouts” and linked them to measurable traits.[5] Research from here can change how the world understands gene function.

In rare diseases a single case report may be the only evidence a doctor elsewhere has to go on. An unverified variant or a doctored image can harm families who have

nowhere else to turn. For us, integrity is everything.

Our research output has grown quickly, and much of that growth is honest hard work. But promotion rules that count papers instead of weighing them have pushed some towards predatory journals,[6] honorary authorship, duplicate publication and plagiarism. Artificial intelligence adds a new temptation. Used openly it helps; used covertly it invents data and references. AI cannot be an author, and its use must be disclosed.[7] Add thin funding, few genetic counsellors, patchy access to sequencing and ethics committees unprepared for genomic studies. JHARG cannot fix all of this, but it will hold every manuscript to internationally accepted standards, set out in our instructions to authors and reviewers.

Research is a skill, and like any clinical skill it has to be taught. We urge everyone, from undergraduate to professor, to train formally in methodology, biostatistics, ethics and scientific writing. Training works. Case-based ethics teaching improves ethical decision-making.[8] In the SORT IT programme, 197 of 228 submitted papers were published or in press, and many went on to shape health policy.[9] Writing programmes in low- and middle-income countries succeeded where teaching was paired with mentorship.[10]

We want JHARG to be the first scientific home of students, residents and early-career scientists. A well-documented case report is an ideal first paper, and where a manuscript has merit our reviewers will explain how to improve it rather than simply reject it. We ask senior colleagues to be mentors, not just names on a byline; mentoring is a proven driver of research productivity.[11] SNARE and JHARG will also explore workshops on methodology, ethics, case-report writing and variant interpretation.

JHARG is also SNARE's scientific voice. The society brings together clinicians, geneticists, laboratory scientists and families, who together can build what Pakistan still lacks: patient registries, shared diagnostic pathways and international links. In time, we hope these pages become a searchable record of rare diseases in our part of the world.

Now picture it grown

I have spent the first part of this editorial on gloom, and I meant every word of it. Let me now tell you what I see when I allow myself to hope, because I mean that just as much.

I see a resident in Multan, Quetta or Gilgit faced with a child whose syndrome nobody on the ward has seen before, who opens JHARG and finds that three Pakistani colleagues saw it first and wrote it down properly. I see a nervous house officer's first case report, made better by a reviewer who took the trouble to explain, cited years later by a geneticist on another continent. I see a registry quietly growing out of these pages. I see families who were once told that nothing is known about their child's condition discovering that something is now known, and that some of it was learned here. I see JHARG indexed, read and trusted, and young researchers in this region choosing it over a predatory journal simply because it is the better home for their work. None of this is a fantasy. Every journal that is respected today was once a thin first issue that very few people believed in.

What it needs is not much, and it needs it from you. It needs your manuscripts, your original research, case reports, reviews and ideas, and your best work rather than the paper everyone else has turned down. It needs experienced colleagues on its panel of reviewers, giving a few honest hours a year and giving them on time. It needs to be cited when it deserves citing, mentioned to students, forwarded to colleagues and brought up at departmental meetings. It needs institutions, here and abroad, to stand behind it with something more than a letter of approval.

Most of all it needs owners. A journal that belongs to everyone on paper and to nobody in practice will die quietly, the way those daughters did. What ended that custom in Arabia was not one grieving mother. It was a whole community learning to see its daughters differently, as a blessing to be protected rather than a burden to be disposed of. That is exactly what will save JHARG. So take it home. Feel protective of it. Let it feel safe in your hands. Complain when it falls short, and then roll up your sleeves and help fix it. If every member of SNARE did just one thing for this journal in the coming year, one paper, one review, one student pointed in its direction, this orphan would have a family, and that family would transform it beyond anything I can manage alone.

Rare does not mean unimportant. Every patient with a rare disease deserves research that is honest, careful and shared. That is the standard we set for JHARG in this first issue. Help us meet it, and hold us to it.

Conflict of Interest None

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