

EpiGene

**Revolutionizing Pediatric Epilepsy Care through Precision
Medicine and Digital Healthcare Technology**



Education

MD degree from King Abdulaziz University.

1

2

Medical Training

Pediatric board residency

Fellowship in pediatric neurology.

Completing fellowship in epilepsy and EEG at the University of Calgary.

3

Research

Focus on genetic and developmental epileptic encephalopathies.

Hanin A. Alsini

Pediatrician, Pediatric Neurologist, and Epilepsy & EEG Fellow

King Fahad Armed Forces Hospital, Jeddah, SA



Agenda



Diverse Nature



Precision Medicine



Sarah's Story



**Bridging Research &
Practice**



**Feasible &
Sustainable**



Cost-Benefit



**The Transformative
Impact**



The Vision Ahead





The Current Epilepsy Landscape

Statistics

- **50 million** people globally.
- **6.5 per 1000** in Saudi children.
- **The highest in the first year of life.**

Challenges

- **Poor seizure control**
- **Medication side effects**
- **Emotional & financial impact of epilepsy.**

Ahmed Al Rumayyan, Neuroepidemiology 2023;57:78-89, DOI: 10.1159/000522442
Bashiri, Genotype-Phenotype Analysis of Children with Epilepsy. Children 2023,



Genetic Foundation of Epilepsy - The New Era

Genetics



Genetic causes were identified in **22%** of epilepsies globally. In Saudi Arabia, **13%** of childhood epilepsy has genetic etiology.

Culture



Higher **consanguinity** rates may indicate a higher contribution of abnormal genes to childhood epilepsy in Saudi Arabia

Targeted Approach



>200 genetic variants, unique treatment pathways tailored to each genetic factor.



The Problem of One-Size-Fits-All

One Size Fits All

- Conventional pharmacotherapy is not effective for everyone.
- Mixed results due to the diverse nature of the disorder.

Disease's Nature

- Epilepsy manifests similarly through the lens of seizures.
- The underlying Genes and their implications are varied.



The Solution: Personalized Medicine



Precision Medicine (PM)

Integration of genetic information, lifestyle factors, & environmental data to tailor therapeutic approach to the patient's needs.



The Impact

Targeted therapies that improve control, reduce side effects, & enhance quality of life.



Evolving Landscape

Transition from a one-size-fits-all approach to a patient-centric model of care.



Sarah: A Beacon of Hope for Precision Epilepsy Care

At one year, Sarah's life has been profoundly impacted by a childhood epilepsy diagnosis.

Her family's journey is mirrored by many others seeking a better path forward.

Let's meet Sarah



Sarah's Story:

Dec 2022

Infantile Seizures

- Born in Alhasa
- Battling unpredictable & debilitating seizures.

Feb 2023

Intractability

- Admissions, ER visits, & PICU with seizures.
- Hyperactivity & aggression.

Feb 2024

Diagnosis

- At KFAFH, presented with seizures.
- A rare, recently discovered X-linked DEE suspected.

March 2024

PCDH19

- NAGS revealed a specific variant mutation in *PCDH19*.
- Successful treatment and seizure control.



Transforming Epilepsy Care through Precision Medicine



Genetic Breakthroughs

The diverse genetic underpinnings of epilepsy, offer the potential to tailor treatments to each individual's unique genetic profile.



Bridging the Gap

While this knowledge holds the key to transforming lives, the insights remain scattered across numerous publications, often inaccessible to busy clinicians.



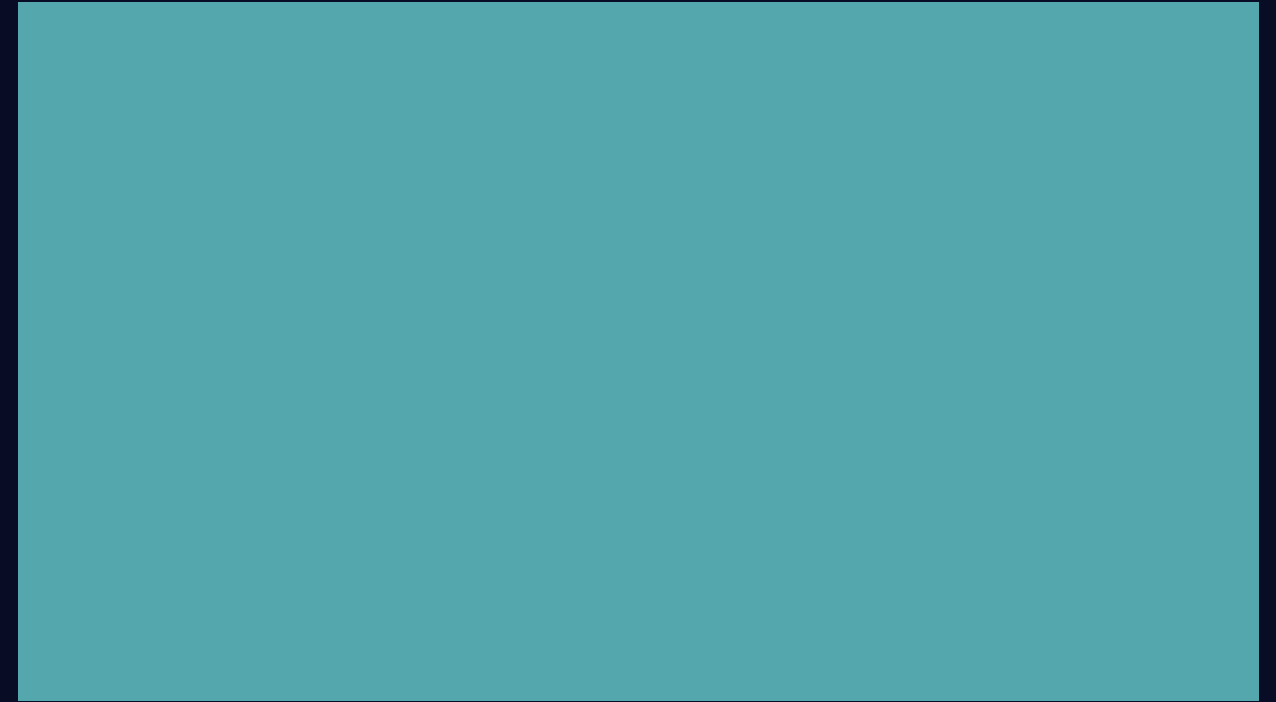
Personalized Approach

Deliver the right treatment to the right patient at the right time, improving outcomes for patients.



EpiGene,

A groundbreaking application that bridges the gap between **genetic research & clinical practice.**



By harnessing the power of digitalization and precision medicine, EpiGene empowers healthcare professionals with instant access to the latest genetic insights & treatment recommendations for epilepsy.



Describing EpiGene

What is the problem?

- Genetic epilepsy management is hindered by the dispersion of crucial genetic information across numerous scientific publications.
- It is challenging for clinicians to access & apply this knowledge in real-time patient care.



What does EpiGene provide?

- Systematic literature review and data extraction
- Comprehensive genetic epilepsy database
- User-friendly app accessible on various devices
- Integration the latest research findings into clinical decision-making with level of evidence



Imagine Ahmed and Fatima's relief when Sarah's doctor, armed with EpiGene, quickly identifies her specific genetic variant early in the disease course and prescribes a targeted treatment that dramatically reduces her seizures and prevents neurocognitive sequelae.

This is not just a hypothetical scenario – it's the future that EpiGene is making possible for countless families across Saudi Arabia and beyond.



Sustainability: Support Structures & Management

(1) **Collaboration** with epilepsy centers & genetic research institutions.

(2) Regular **updates** & version control systems.

(3) User feedback **integration** & expert review panels.

(4) Regular training & update sessions for **users**.

(5) **Workshops** & online courses for healthcare professionals.

(6) Continuous **engagement** with healthcare professionals & patients.



Novelty: Unique Approach

No evidence of a comparable platform that combines;

A comprehensive database of **genetic variants** associated with epilepsy syndromes

Real-time integration of the latest research findings

A user-friendly, smartphone-compatible application for point-of-care use

While existing platforms like the **GET (Gene, Epilepsy, Treatment) website** focus on integrating genetic information into EMR, **EpiGene** takes this concept further by offering **immediate, portable access to this crucial information across various devices.**



Approach: Development & implementation

Plan

1. Systematic literature review & data extraction
2. Database design & structure
3. Application roadmap
4. User interface design based on clinician feedback.

Execute

1. Phased development approach, starting with core functionality
2. Beta testing with select epilepsy centers
3. Iterative improvements based on user feedback

Test

1. Rigorous quality assurance protocols
2. Validation of genetic information accuracy
3. User testing in clinical settings
4. Performance testing for various devices and operating systems

Expected Results

1. Improved speed & accuracy in diagnosis
2. Enhanced treatment efficacy through PM
3. Increased clinician confidence in genetic data interpretation
4. Streamlined clinical workflows in epilepsy management



Leveraging existing resources & technologies;

Genetic Databases

Utilizes established
databases e.g.
**MEDLINE,
Genecards, &
MedlinePlusGenetics.**

Software Development:

Employs available
cross-platform
development
frameworks.

Cloud Infrastructure

Utilizes scalable cloud
services for **data
storage & processing**

The EpiGene project, while not yet implemented, demonstrates feasibility



Designed with interoperability & ease of adoption:

Complementary

Standalone app
with potential for future
EHR integration

User-friendly

Intuitive design
requiring training for
healthcare professionals

Flexible

On personal devices or
institution-provided
hardware

Scalable

Easy updates &
additions of new
genetic information

Customizable features

Ability customize to specific institutional needs or regional genetic profiles



EpiGene: the economic viability

SR 450K

Development Costs

SR 150K

Database Creation and Population

SR 80K

UI Design and Testing

SR 100K

Annual Software Maintenance

SR 45K

Database Curation and Updates

SR 35K

Cloud Hosting and Infrastructure

SR 150K

Technical Support Team

SR 250K

Genetic Research Team

SR 50K

Initial Marketing Campaign

SR 50K

Ongoing Marketing Efforts

SR 50K

Training Programs

SR 50K

Regulatory Compliance



EpiGene Benefits

SR 500K

Healthcare Cost Savings

per patient from reduced tests and admissions

SR 100K

Practitioner Subscriptions

SR 250K

Data Insights for Research

SR 500K

Healthcare Institution Licensing Fees

1

Improved Patient Outcomes

Enhanced quality of life and reduced time from diagnosis to effective treatment

2

Healthcare Professional Satisfaction

Increased confidence in treatment and reduced burnout

3

Advancement of Epilepsy Research

Accelerate research in genetic epilepsies and new treatments

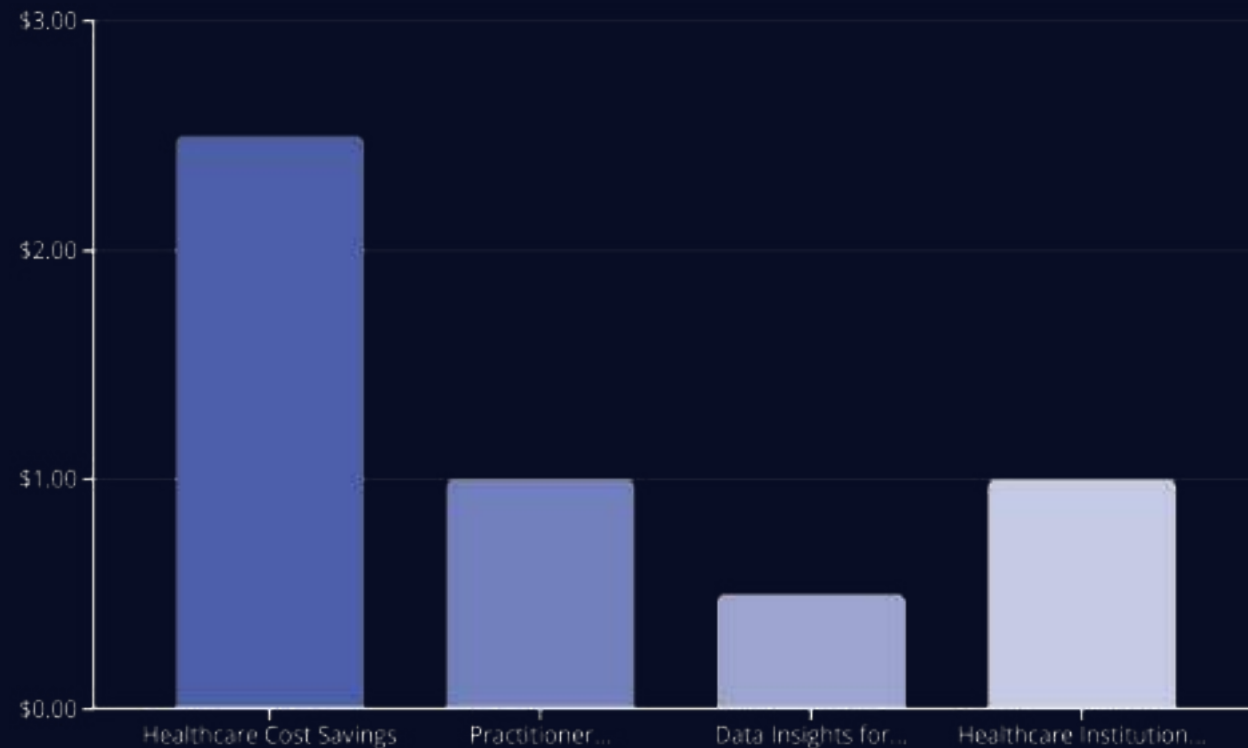
4

Enhanced Reputation

Positioning institutions as leaders in epilepsy care



Cost-Benefit Summary



Positive return on investment, financially & societally.

The potential for transforming epilepsy care, in a region with high prevalence of genetic epilepsies underscores the project's value proposition.



Impact Assessment: Transforming Lives

Beneficiaries

- Patients
- Neurologists & Epileptologists
- Genetic Counselors

1ry

- Families of patients
- General Practitioners
- The Healthcare system

2ry



Key Outcomes

Improved Diagnostic Accuracy



Enhanced Treatment Efficacy

Streamlined Clinical Workflow



Patient Empowerment

Advancement of Epilepsy Research



Patient Safety Risks

1. Data Privacy and Security

2. Misinterpretation of Genetic Information

3. Treatment Errors

4. Overconfidence in Technology



Ensuring the Safety & Reliability

1. Data Protection

2. Information Accuracy

3. Clinical Decision Support

4. User Training

5. Feedback and Monitoring



EpiGene



support, global registry recruitment, variant interpretation, data visualization, and collaboration opportunities with researchers and clinicians.

Gene Analysis



An overhead view of a classroom with several children sitting at white desks. They are using various digital devices: a laptop, a tablet, and a smartphone. The desks are arranged in a grid-like pattern on a light-colored wooden floor. The children are wearing colorful clothing, and the overall atmosphere is one of focused learning and technology use.

As we delve deeper into the EpiGene project, remember Sarah and the thousands of children like her..

Their stories drive our mission to revolutionize epilepsy care through the seamless integration of digitalization and precision medicine.





With EpiGene, we're not just treating seizures; we're transforming lives, **one genetic profile at a time.**





With EpiGene, we're not just treating seizures; we're transforming lives, **one genetic profile at a time.**

