EpiGene

Revolutionizing Pediatric Epilepsy Care through Precision Medicine and Digital Healthcare Technology

Education

1

MD degree from King Abdulaziz University.

2

Medical Training

Pediatric board residency

Fellowship in pediatric neurology.

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

Research

3

Focus on genetic and developmental epileptic encephalopathies.

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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

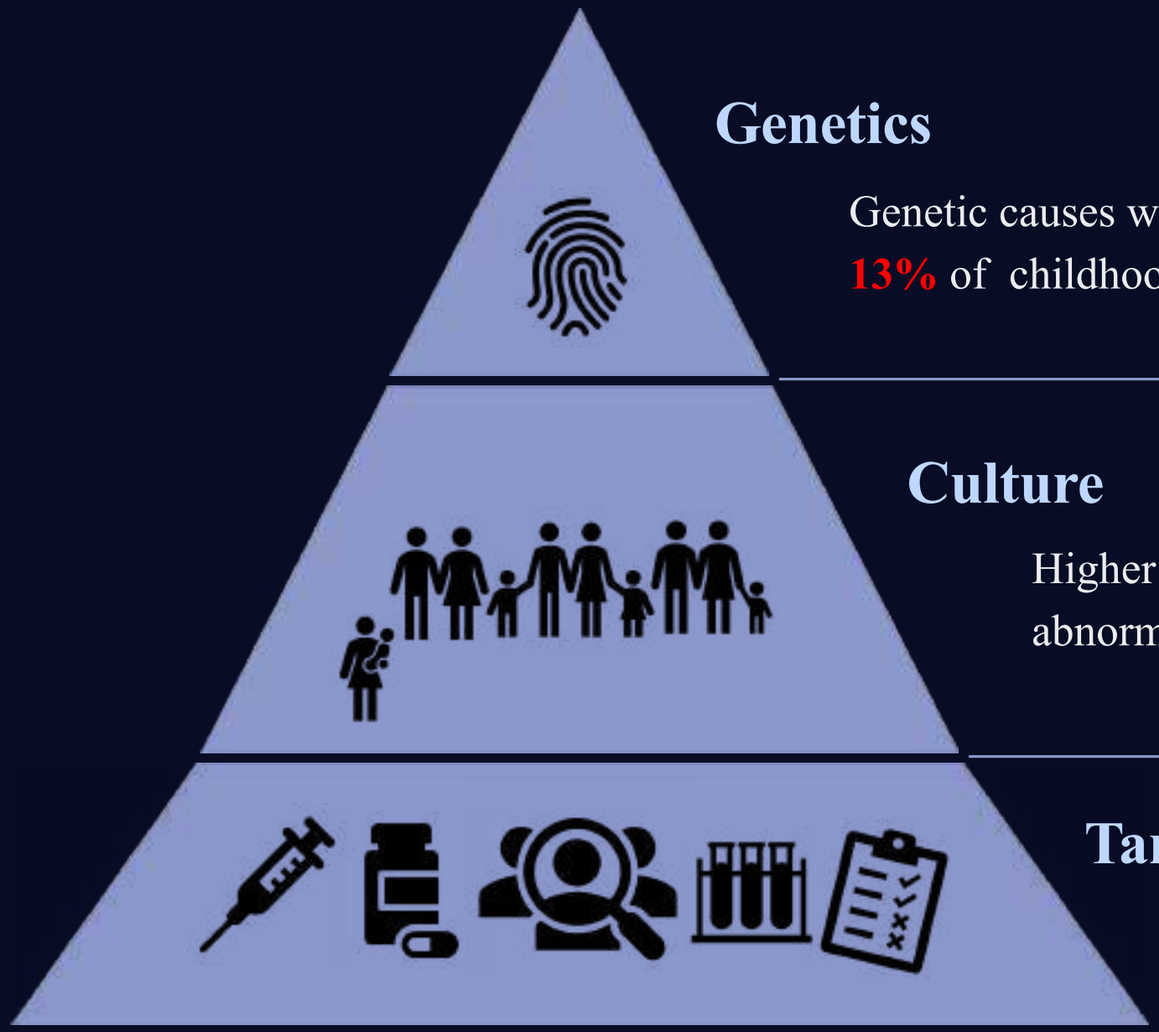
- **50 million** people globally.
- **6.5 per 1000** in Saudi children.
- The **highest in the first year of life.**

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.

Bashiri, Genotype–Phenotype Analysis of Children with Epilepsy. Children 2023, 10,1334. https://doi.org/10.3390/ children10081334

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

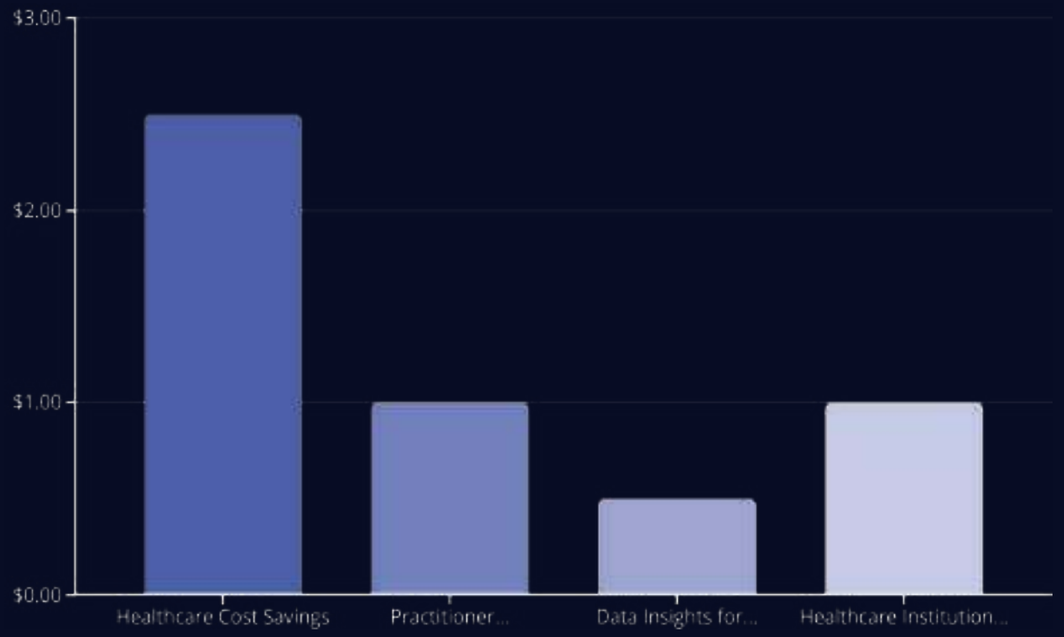
3 **Advancement of Epilepsy Research**

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

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.

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