FRAGILE X INTERNATIONAL

Fragile X Premutation Associated Conditions (FXPAC)

Kirsten Johnson[1*], Jonathan Herring[2] and Jörg Richstein[3]

[1] The Fragile X Society, Great Dunmow, United Kingdom
[2] Faculty of Law, University of Oxford, Oxford, United Kingdom
[3] Interessengemeinschaft Fragiles-X e.V, Rostock, Germany

The European Fragile X Network (EFXN) proposes that Fragile X Premutation Associated Conditions (FXPAC) be adopted as a universal term covering any condition linked to the Fragile X premutation. To date, there has not been an umbrella term assigned to issues associated with the FMR1 premutation, though several defined conditions which affect some premutation carriers, namely Fragile X-associated Primary Ovarian Insufficiency (FXPOI) and Fragile X-associated Tremor/Ataxia Syndrome (FXTAS), are now commonly accepted. An overarching term covering all FX premutation conditions will help doctors in determining how the premutation might be affecting their patient; and encourage researchers to explore the interrelationships of the various conditions affecting premutation carriers. Further, there are ongoing discoveries about physical and psychological issues faced by premutation carriers, and a new term helps encompass all of these burgeoning developments.

Frontiers in Pediatrics May 27, 2020

FXPAC

Cells. 2022 Mar 19;11(6):1044. doi: 10.3390/cells11061044.

The Use of "Retardation" in FRAXA, FMRP, FMR1 and Other Designations

Jonathan Herring [1], Kirsten Johnson [2 3], Jörg Richstein [3 4]

Affiliations + expand

PMID: 35326495 PMCID: PMC8947541 DOI: 10.3390/cells11061044

Free PMC article

Abstract

The European Fragile X Network met in Wroclaw, Poland, November 2021, and agreed to work towards the eradication of the word "retardation" in regard to the naming of the fragile X gene (FRAXA) and protein (FMRP). There are further genes which have "retardation" or abbreviations for "retardation" in their names or full designations, including FMR1, FMR2, FXR1, FXR2, NUFIP1, AFF1, CYFIP1, etc. "Retardation" was commonly used as a term in years past, but now any reference, even in an abbreviation, is offensive. This article discusses the stigmatisation associated with "retardation", which leads to discrimination; the inaccuracy of using "retardation" in these designations; and the breadth of fragile X syndrome being beyond that of neurodiversity. A more inclusive terminology is called for, one which ceases to use any reference to "retardation". Precedents for offensive gene names being altered is set out. The proposal is to approach the HGNC (HUGO [Human Genome Organisation] Gene Nomenclature Committee) for new terminology to be enacted. Ideas from other researchers in the field are welcomed.

Keywords: FMR1; FMR2; FMRP; FRAXA; FXR1; FXR2; fragile X premutation associated conditions (FXPAC); fragile X syndrome (FXS).

Neurodiversity
Volume 2, 2024

https://doi.org/10.1177/27546330241287685

SageJournals

Commentary

The joys of fragile X: Understanding the strengths of fragile X and delivering a diagnosis in a helpful, holistic way

Jonathan Herring [1,2], Kirsten Johnson [2], Gaia Scerif[3,4], Emilie Weight[5], Jörg Richstein[2], Hayley Crawford[4,6], Heloise Robinson[7], Radhini Gawarammana [7], and Katherine Ellis[4,8,9]

The purpose of this article is to provide a positive framework for a doctor, geneticist, genetic counsellor or other professional to deliver a fragile X syndrome diagnosis and to offer support thereafter. Our aim is not to glamourise the condition nor downplay its impacts but rather to suggest that a more accurate and holistic definition of fragile X syndrome would cover all the aspects of the condition, including its variability and positive aspects. Fragile X syndrome is commonly described in terms of the 'problems' associated with it and stigmatic language is used. Instead, we believe that giving diagnosis and support in a holistic and family-centred way is imperative. In delivering a fragile X syndrome diagnosis, the strengths of those living with fragile X syndrome should be the starting point, encouraging development that builds on those strengths while supporting the areas of need. This article will set out these more positive aspects of fragile X syndrome to assist those providing a diagnosis of fragile X syndrome.

Lay abstract The purpose of this article is to provide a positive framework for a doctor, geneticist, genetic counsellor or other professional to deliver a fragile X syndrome (FXS) diagnosis and offer support. Our aim is to give a more accurate and holistic definition of FXS. This would cover all the aspects of the condition, including its variability and positive aspects. FXS is commonly described in terms of the 'problems' associated with it and stigmatic language is used. Instead, we believe that giving diagnosis and support in a holistic and family-centred way is important. In delivering a FXS diagnosis, the strengths of those living with FXS should be the starting point. These strengths should be built on at the same time as supporting areas of need.

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Homage to Prof. Alessandra Murgia

1st Fragile X International Congress

FRAGILE X INTERNATIONAL

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BARCELONA, SPAIN
7-8 NOVEMBER, 2024

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Volume 20 Supplement 1

Abstracts of the 1st Fragile X International Congress

Meeting abstracts | Open access | Published: 24 April 2025

Meeting Abstracts of the 1st Fragile X International Congress

Orphanet Journal of Rare Diseases **20**, Article number: 173 (2025) | Cite this article

620 Accesses | Metrics

Editors: Professor Gaia Scerif (1) and Dr Kirsten Johnson (2)

1. Professor of Developmental Cognitive Neuroscience, University of Oxford and St. Catherine's College, Department of Experimental Psychology, Anna Watts Building, OX2 6GG, Oxford, UK
2. President, Fragile X International, 11 Rue d'Egmont, 1000-Bruxelles, Belgium

Fragile X Syndrome (FXS) Guideline Development Overview

Kirsten Johnson, Fragile X International

Fragile X International Workshop, Padua 20.06.2025

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Stages of the Guideline Development Process

Stage 1

Preparation

- Prioritisation
- Stakeholder Engagement
- Select Topic
- External Consultation on topic
- Forming Guideline Development Group (GDG)
- Determining Scope
- Development of PICO Questions

Stage 2

Research

- Draft Systematic Review Protocol
- Search, screening and selection relevant papers
- Data extract for systematic review (GRADE)

And/Or

- Summarising key evidence using GRADE
- Consensus building (DELPHI)

Stage 3

Writing

- Evidence to decision
- Formulate Recommendations
- Writing Guideline
- Peer review
- Finalize Guideline

Stage 4

Implementation

- Publication in scientific journal
- Production of the guideline
- Communications plan
- Develop lay-persons version
- Launch event
- Dissemination with the patient community

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Governance Structure

Guideline Development Group (x35, 12 MSs)

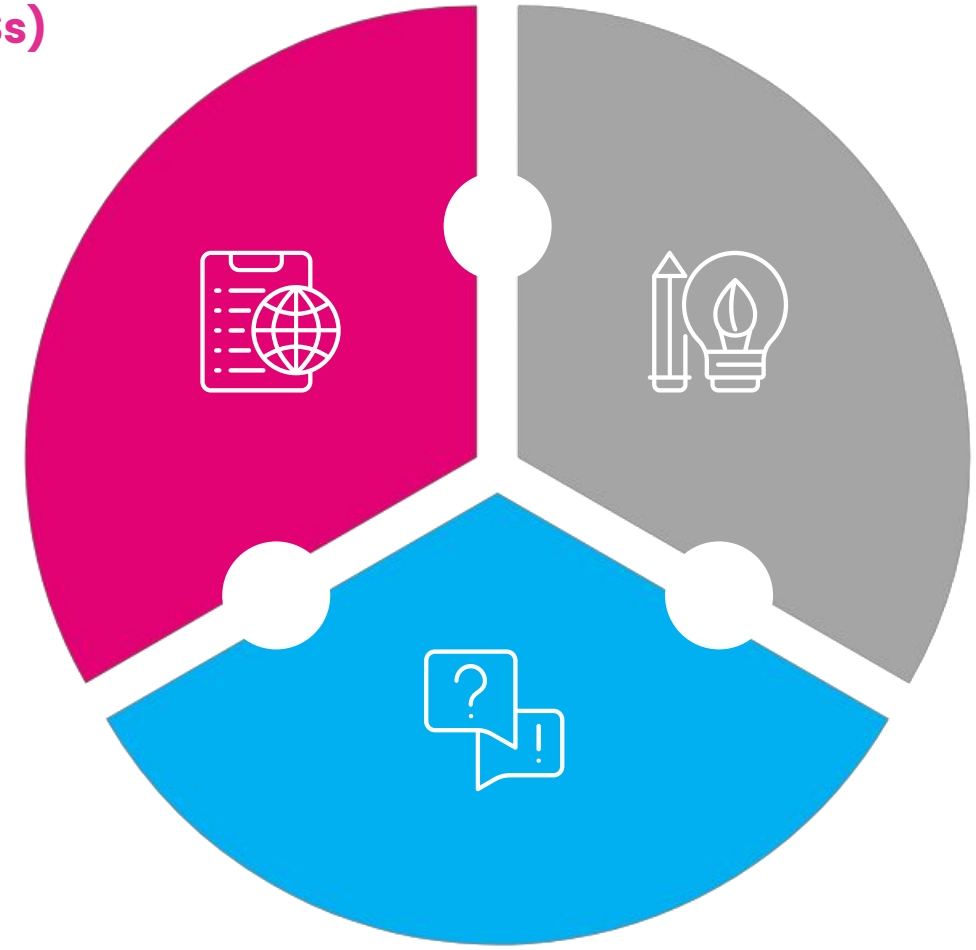
GDG is a **multidisciplinary** group responsible for reviewing, updating, and publishing FXS guidelines; advising on scope; evaluating evidence using GRADE; achieving consensus through evidence-based methods; reviewing lay information and engaging stakeholders.

Core Writing Group (x12 Leads, 9 MSs)

The CWG, a subset of the GDG, serves as its operational arm, engaging with the GDG for input on guideline scope, research, and drafting recommendations. The CWG will consist of 8–12 experts from 4–5 countries to form the CWG.

Lived Experience Advisory Group

The LEAG will advise on guideline scope, target population, and key questions. The group will assess evidence, review recommendations, and gather community input to ensure patient perspectives inform the guideline development.



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Core Writing Group

Lead	Speciality	Institution	Country
Bram DIERCKX	Child and Adolescent Psychiatrist	Erasmus Medical Centre	The Netherlands
Elisa DI GIORGIO	Neuropsychologist	University of Padua	Italy
David GODLER	Geneticist	University of Melbourne	Australia
Frank KOOY	Medical Geneticist	University of Antwerp	Belgium
Kirsten JOHNSON (co-chair)	Expert by Lived Experience	Fragile X International	UK
Caterina PRIOR	Neurodevelopmental Paediatrician	Northern Maternal & Child Centre, Local Health Unit of Santo António	Portugal
Ana ROCHE	Paediatric Neurologist	University Hospital Vall d'Hebron	Spain
Bitten SCHOENEWOLF-GREULICH	Clinical Geneticist	Rigshospitalet	Denmark
Gaia SCERIF	Psychologist	University of Oxford	UK
Zimi SAWACHA	Movement Specialist & Occupational Therapy	University of Padova	Italy
Andrew STANFIELD (chair)	Consultant Psychiatrist	University of Edinburgh Patrick Wild Centre	UK
Matt BOLZ-JOHNSON	Project Manager	Fragile X International	Germany

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Stage 1: Preparation (Completed)

Preparation Tasks:

- **Select topics** for the guideline e.g. priority-setting survey
- Select **Chair & Core Writing Group**
- **Determine the guideline scope and clinical questions** incl. PICO questions (**P**opulation, **I**ntervention, **C**omparison & **O**utcomes)
- Selection of members and set up the Guideline Development Group (GDG)
- Engagement with external stakeholders on the scope

Patient Involvement:

- **Patient/representatives as core members** in the Core writing Group and in the Guideline Development Group (1-2 patients/representatives)
- **Set up a Lived Experience Advisory Group** to get wider patient perspectives (8-10 patients/representatives)
- Patients should **advise on the guideline scope**: and suggest aspects important to them e.g.: PICO questions
- **Rate the importance of outcomes** from patient perspective

- The GDG Chair is responsible to support effective patients and carers involvement
- Project Manager set out at beginning where patients will be involved, with timelines
- Patients involved should receive information, support and training on the guideline development process and methodology.

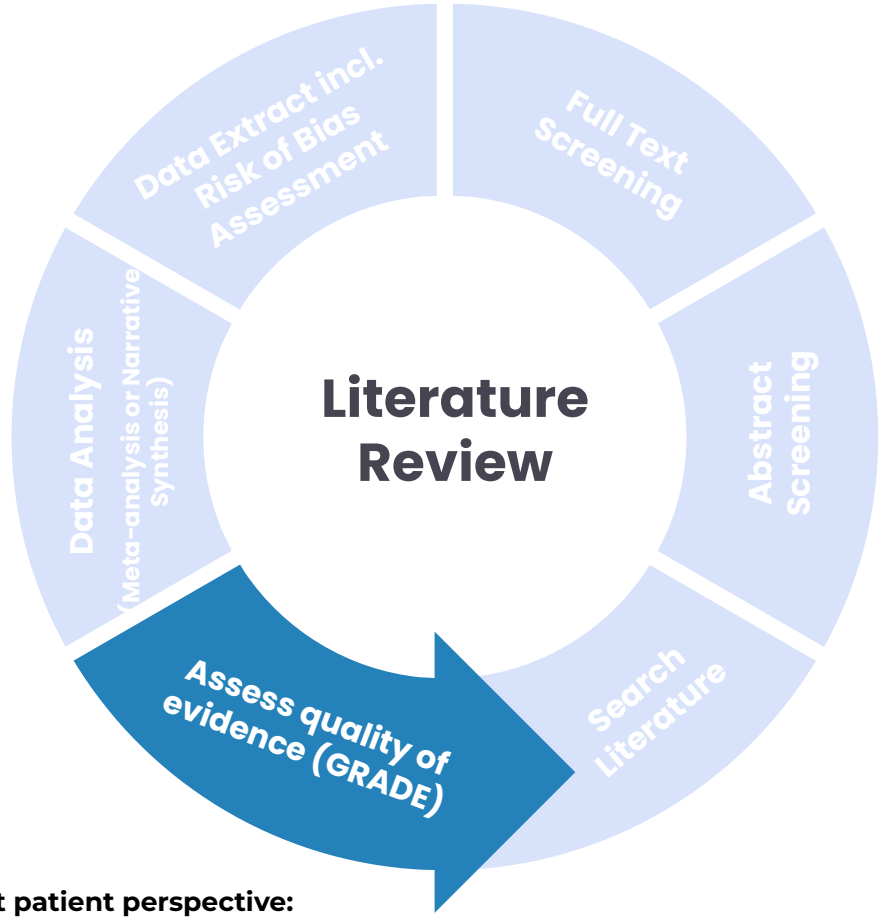
Stage 2: Research (Q2 2025)

Systematic Review Protocol

Guideline Scope (PICO)

Formulate a clear, well-designed research question

Writing a systematic review protocol



Selection & Screening:

- **All GDG members** review titles, abstracts and full text articles.
- **Identify important publications** for inclusion in systematic review
- Initial screening completed by technical team.
- Final screening to be conducted by **all GDG members** against inclusion and exclusion criteria. Any doubts on include to be discussed by the GDG.

Guideline Development Group (GDG)

Appraisal & synthesis of evidence:

- Identify gaps in evidence and gather **unpublished non-experimental data** (expert-based evidence)
- **Identify indirect evidence for consideration.**
- **Identify the evidence underlying their opinions** and **judge it's quality**
- **Discuss and grade evidence** for risk of bias, inconsistency, indirectness, imprecision and publication bias (using GRADE)

TOP TIPS

Can consider other ways to get patient perspective:

- Review of patient experiences via published qualitative literature. e.g.: focus group, interviews about experience of diagnosis. This can give insight and suggest areas of good practice.
- Where no qualitative literature exists, conduct additional research e.g.: multi-lingual survey or consult the Lived Experience Advisory Group about specific aspects.

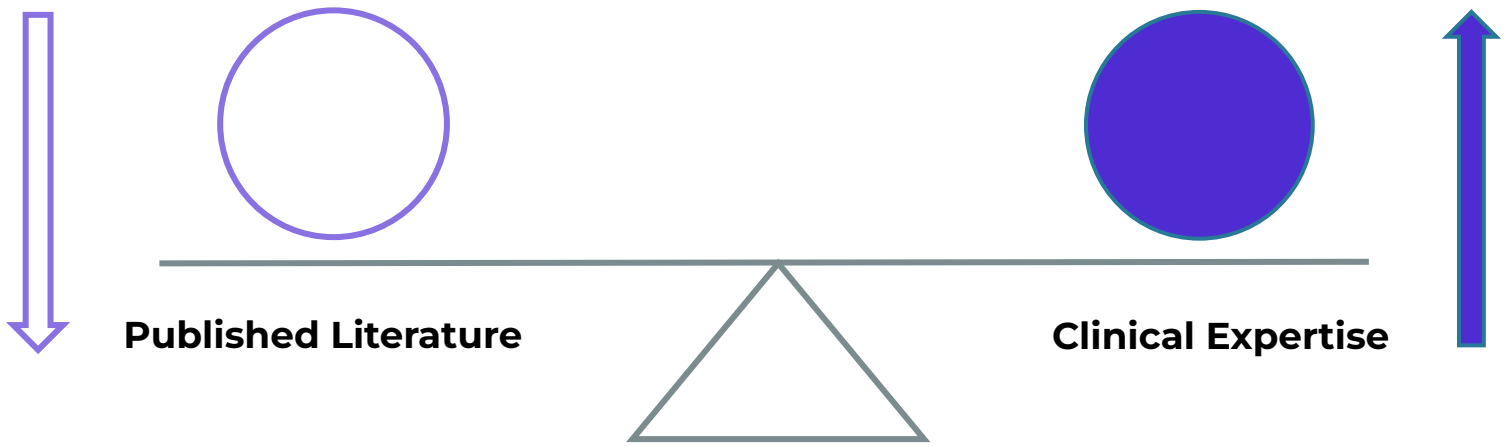
Enhanced GRADE Approach

Systematic Review using GRADE

- Clinical practice guidelines are based on a systematic review of the evidence.
- The level of evidence needs to be stated and graded using GRADE.

Consensus Building using DELPHI

- Clinical consensus statements are developed where evidence is limited or lacking to reflect opinions drafted by experts
- Draw on the wealth of clinical experience to improve quality of care



Enhanced GRADE Approach

- Evaluation of evidence for rare disease guidelines requires an 'enhanced' GRADE approach with DELPHI
- Literature review and consensus building approach.

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Consensus Building (Q3 2025)

Preparation

- Participate in formal consensus-building procedures where there are gaps in evidence.
- Consensus Panel (at least 10-30 participants) must be multidisciplinary **incl. care professionals, healthcare managers & patients/carers.**

1

Step 1: Build Recommendations

- GDG draft recommendations after the literature review and grading phase.
- **Recommendations with low quality evidence or conflicting evidence are selected to proceed in the Delphi procedure.**

2

Step 2: Consensus Exercise

- First round of an online survey: 4-point scale to agree/disagree.
- Second round includes the overall rating of each statement as well as the participants own rating.
- Third round (when needed).

3

Step 3: Consensus Meeting

- virtual consensus meeting to discuss the results of the survey and address any significant variations.
- Possible re-voting is necessary in areas where no consensus was obtained after two rounds.

4

Step 4: Recommendations

- Draft final recommendations

TOP TIPS

- Patients involved can identify experts to be included in the Delphi process.
- Patients should be included in the Consensus Panel

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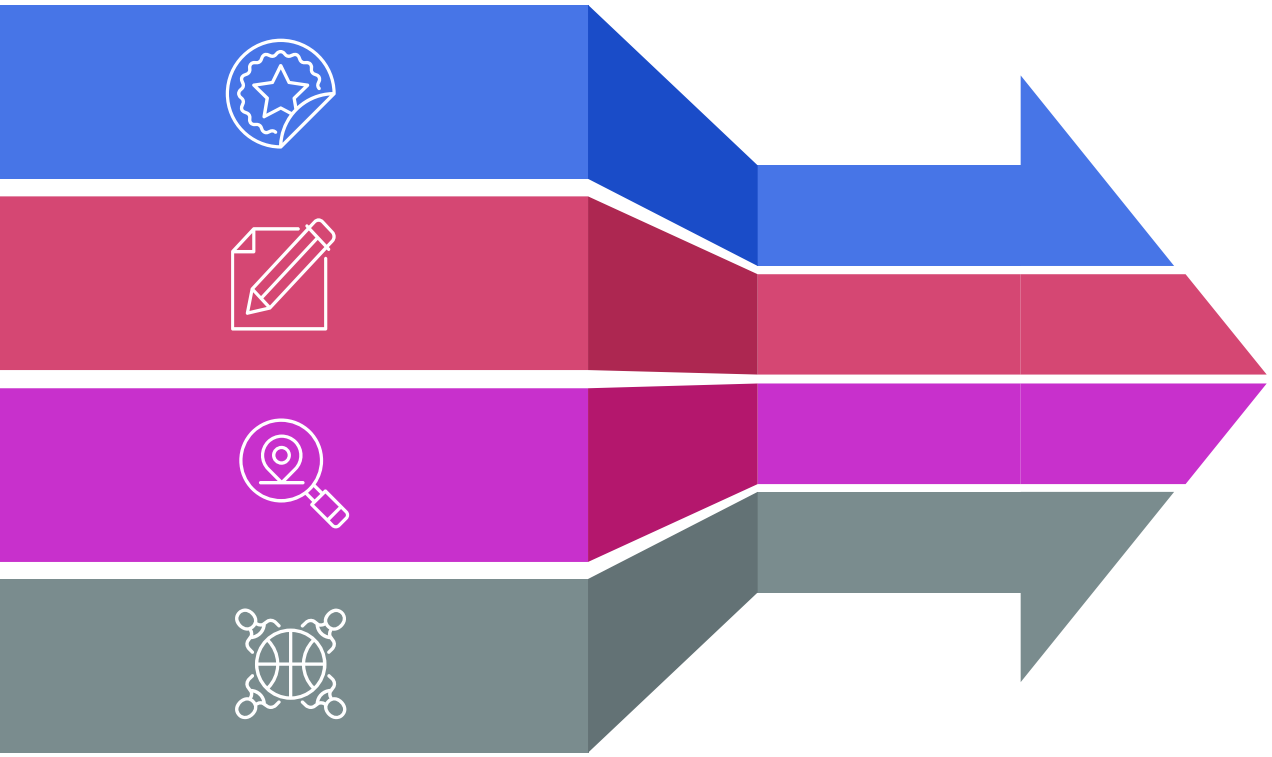
Stage 3: Writing (Q4 2025)



EVIDENCE TO DECISION

E2D Meeting make judgements for each outcome for which there is a desirable effect, taking into account the value that patients place on each outcome.

Consider to what extent are patients willing to accept the possibility of adverse effects against a favourable clinical outcomes.

DEVELOP RECOMMENDATIONS

Professional members write the guideline.

Patients can add valuable insight into how the recommendations can work in practice, which is essential here to make sure final guidelines are useful in the real world.

IDENTIFY GAPS

Evidence gaps often in areas patients consider important e.g.: pain and discomfort.

These areas often have no research evidence from the main lit review – so patient perspectives are vital.

EXTERNAL REVIEW & PUBLICATION

Recommended that at least 2-6 reviewers (of which one at least should be patients and carers) are engaged in the process.

Patients should be included as co-authors or noted in the acknowledgements.

TOP TIPS

- Secure a small budget to support the patient representatives in the LEAG to develop a plain language summary.
- Start drafting a plain language summary in parallel to writing the clinical guideline.

Progress Update

FXS Guideline(s)

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Fragile X Syndrome (FXS): **Guideline Scope**

Screening & Diagnosis #1

Covers when and how to screen or test for Fragile X Syndrome (FXS), including:

- Routine, reproductive, and newborn screening
- Diagnostic criteria by age and sex
- Genetic testing methods (e.g., repeat length, methylation)
- Communicating diagnoses and providing support
- Cascade testing and family planning guidance

Management, care and support in children and adolescents with FXS #2

Focuses on developmental support across key domains:

- Communication, cognition, motor, and sensory skills
- Physical and mental health conditions
- Behavioural and sleep challenges
- Educational placement and lifestyle needs
- Family support and integrated care coordination

Management, care and support in adults with FXS #3

Addresses lifelong care needs and quality of life:

- Ongoing developmental, physical, and mental health monitoring
- Behaviour and sleep management
- Promoting independence, healthy lifestyle, and relationships
- Support for families and coordinated adult services

FXS Screening & Diagnosis Scope #1

SCREENING

Is routine screening of the general population recommended for FXS?

INDICATIONS FOR DIAGNOSTIC TESTING

What clinical features (red flag symptoms) should prompt a diagnostic test for FXS?

When should prenatal testing for FXS be offered and using what methods?

How should cascade testing be conducted following a diagnosis of FXS in a family member?

When should an older diagnostic test be updated; what are the clinical situations or other factors that would justify this?

TESTING METHODS

What approaches should be taken when testing for FXS?

What approaches should be taken when testing someone for a premutation?

Should X-inactivation be considered when testing females for fragile X or the premutation?

DIAGNOSTIC PROCESS

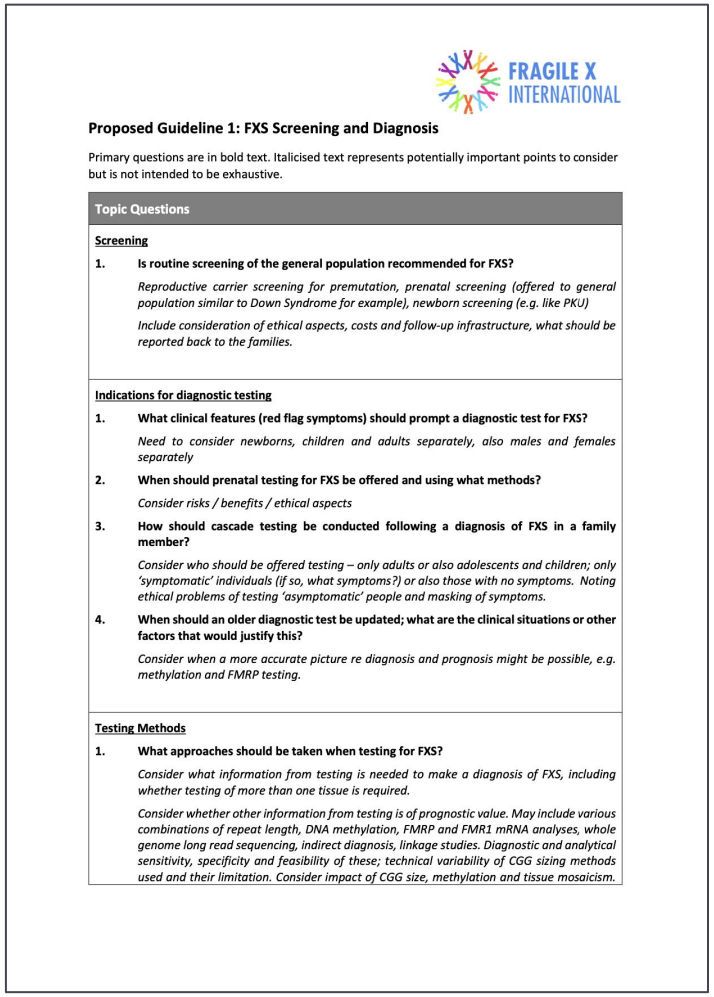
What support is required when FXS is suspected?

How best do we deliver the diagnosis of FXS to individuals and families?

What are the pathways to support, assessment and treatment which need to be in place in the early post-diagnostic period?

To whom, when and how should psycho-social support and genetic counselling be provided, and what information should be provided

#1 FXS Guideline Update



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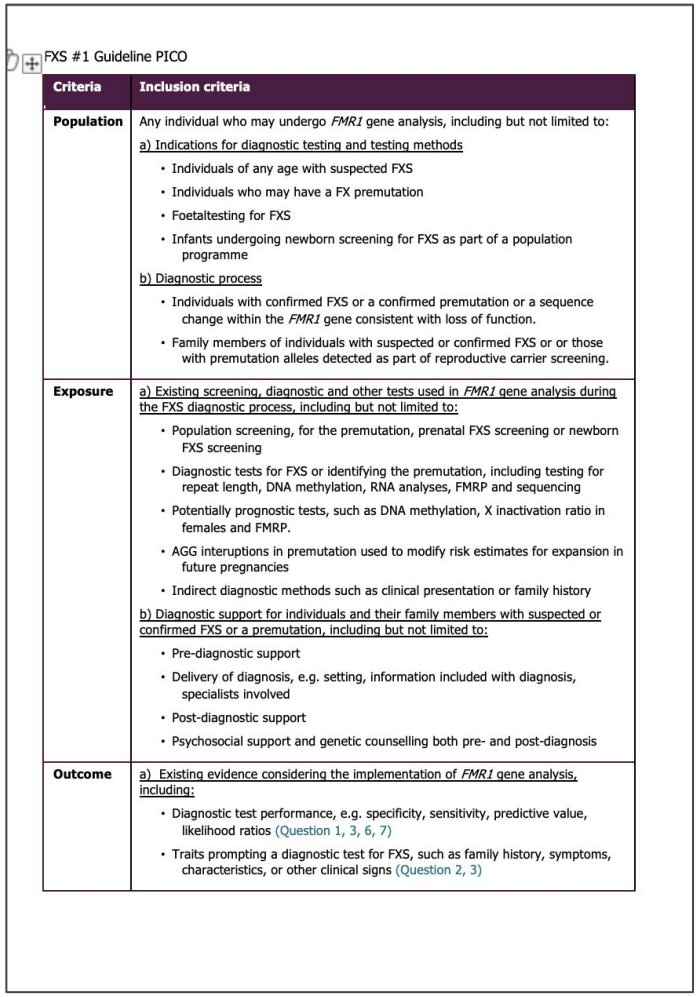
Proposed Guideline 1: FXS Screening and Diagnosis

Primary questions are in bold text. Italicised text represents potentially important points to consider but is not intended to be exhaustive.

Topic Questions
Screening 1. **Is routine screening of the general population recommended for FXS?** *Reproductive carrier screening for premutation, prenatal screening (offered to general population similar to Down Syndrome for example), newborn screening (e.g. like PKU)* *Include consideration of ethical aspects, costs and follow-up infrastructure, what should be reported back to the families.*
Indications for diagnostic testing 1. **What clinical features (red flag symptoms) should prompt a diagnostic test for FXS?** *Need to consider newborns, children and adults separately, also males and females separately* 2. **When should prenatal testing for FXS be offered and using what methods?** *Consider risks / benefits / ethical aspects* 3. **How should cascade testing be conducted following a diagnosis of FXS in a family member?** *Consider who should be offered testing – only adults or also adolescents and children; only 'symptomatic' individuals (if so, what symptoms?) or also those with no symptoms. Noting ethical problems of testing 'asymptomatic' people and masking of symptoms.* 4. **When should an older diagnostic test be updated; what are the clinical situations or other factors that would justify this?** *Consider when a more accurate picture re diagnosis and prognosis might be possible, e.g. methylation and FMRP testing.*
Testing Methods 1. **What approaches should be taken when testing for FXS?** *Consider what information from testing is needed to make a diagnosis of FXS, including whether testing of more than one tissue is required.* *Consider whether other information from testing is of prognostic value. May include various combinations of repeat length, DNA methylation, FMRP and FMR1 mRNA analyses, whole genome long read sequencing, indirect diagnosis, linkage studies. Diagnostic and analytical sensitivity, specificity and feasibility of these; technical variability of CGG sizing methods used and their limitation. Consider impact of CGG size, methylation and tissue mosaicism.*

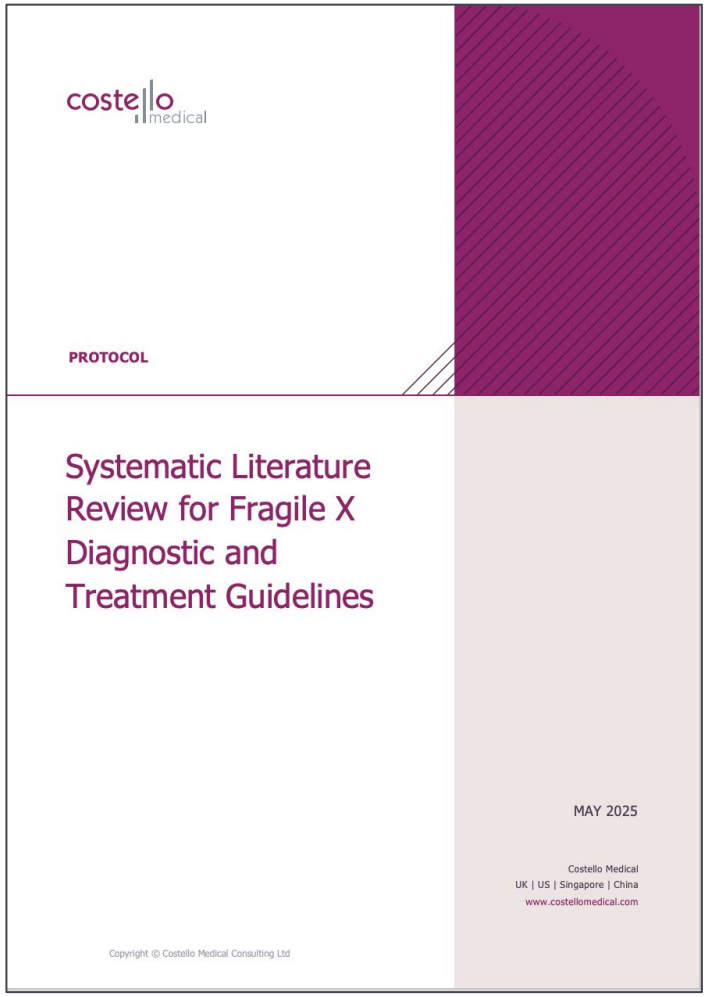
Guideline Scope



FXS #1 Guideline PICO

Criteria	Inclusion criteria
Population	Any individual who may undergo *FMR1* gene analysis, including but not limited to: a) Indications for diagnostic testing and testing methods • Individuals of any age with suspected FXS • Individuals who may have a FX premutation • Foetaltesting for FXS • Infants undergoing newborn screening for FXS as part of a population programme b) Diagnostic process • Individuals with confirmed FXS or a confirmed premutation or a sequence change within the *FMR1* gene consistent with loss of function. • Family members of individuals with suspected or confirmed FXS or or those with premutation alleles detected as part of reproductive carrier screening.
Exposure	a) Existing screening, diagnostic and other tests used in *FMR1* gene analysis during the FXS diagnostic process, including but not limited to: • Population screening, for the premutation, prenatal FXS screening or newborn FXS screening • Diagnostic tests for FXS or identifying the premutation, including testing for repeat length, DNA methylation, RNA analyses, FMRP and sequencing • Potentially prognostic tests, such as DNA methylation, X inactivation ratio in females and FMRP. • AGG interuptions in premutation used to modify risk estimates for expansion in future pregnancies • Indirect diagnostic methods such as clinical presentation or family history b) Diagnostic support for individuals and their family members with suspected or confirmed FXS or a premutation, including but not limited to: • Pre-diagnostic support • Delivery of diagnosis, e.g. setting, information included with diagnosis, specialists involved • Post-diagnostic support • Psychosocial support and genetic counselling both pre- and post-diagnosis
Outcome	a) Existing evidence considering the implementation of *FMR1* gene analysis, including: • Diagnostic test performance, e.g. specificity, sensitivity, predictive value, likelihood ratios (Question 1, 3, 6, 7) • Traits prompting a diagnostic test for FXS, such as family history, symptoms, characteristics, or other clinical signs (Question 2, 3)

PICO Questions



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PROTOCOL

Systematic Literature Review for Fragile X Diagnostic and Treatment Guidelines

MAY 2025

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Systematic Review Protocol

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Guideline Development Timeline

FXS Guideline Development GAANT Chart – Version 1.0

Reference	Tasks	Q1 2025			Q2			Q3			Q4			Q1 2026			Q2			Q3			Q4		
		J	F	M	A	M	J	J	A	S	O	N	D	J	F	M	A	M	J	J	A	S	O	N	D
	Preparation Stage (1)																								
Milestone	**Core Writing Group - KOM held**																								
	• Access to cloud platform																								
	• Set up Guideline Development Group & Core Writing Group																								
	• Set up Lived Experience Group																								
	• Finalised Revised Scope (PICO)																								
	Research Stage (2)																								
	Systematic Review																								
	• Screening publications																								
	• Full paper review																								
	• GRADE Evidence																								
	• Data extraction																								
	• Draft evidence summary(ies)																								
Milestone	Draft initial recommendations & gaps																								
	Consensus Building Exercise																								
	• 1st round: online survey																								
	• 2nd round: online survey																								
Milestone	Final expert consensus meeting held																								
	Writing Stage (3)																								
	• Agree format and style of guideline																								
	• Statement engagement on content																								
	• Draft Guideline																								
	• Draft 'at-a-glance' summary																								
	• Draft Plain Language Summary																								
	• Walk through final guideline with GDG																								
Milestone	Final FXS CPG Completed																								
	Approval																								
	• Assess resource implications for NHS																								
	• Peer review using AGREE II																								
	• Share with EB Stakeholders																								
	• Final revision																								
	Draft publication																								
Milestone	FXS CPG Approved																								

	#1 Guideline		#1 Guideline		#1 Guideline

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Fragile X International Global Federated Registry

+

FraXI Registry

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OVERVIEW

DECENTRA HEALTH

Decentra Health Inc

- Headquarters - New Jersey, USA
  - Offices - Toronto, UK, North Carolina
- Private Company

Our Mission

- Unlock the collective power of patient data to drive groundbreaking research and accelerate the development & delivery of life-saving treatments.

Our Vision

- Create a privacy-preserving, collaborative infrastructure that connects patient data globally to unleash AI-driven research and moves science forward.

Our Solution

- Powered by our breakthrough technology (The DEM), our Decentralized AI platform connects & analyzes patient data globally with complete privacy and control.

Our Executive Team

Dustin O'Dell
Co-founder, CEO

- *Experian Data Services, BD & Strategy*
- *6th employee, BD Lead AdTheorent (IPO, Acquired)*
- *Co-founder Barometric (Acquired)*
- *Co-founder, SymetrML*

Chris Hendriksz MD
Co-founder, Chief Medical Officer

- *Professor, Human Metabolomics, North-West University, Pediatrics Pretoria University, South Africa*
- *Global Clinical Dev Lead, Nestle Health Science (Rare Disease & Innovative Pharmaceuticals)*
- *Inovled In over 20+ drug approvals whilst running rare disease centres in NHS UK*
- *Chief Community Impact Officer A Rare Cause*

Eric Faulkner
Co-founder, Chief Scientific Officer

- *Global VP Novartis GTx (RWE lead for Zolgensma)*
- *VP, PPD (Value, Access & RWE)*
- *Practice Lead, IQVIA*
- *Involved In over 50+ drug approvals*

OUR FEDERATED AI PLATFORM

Decentra Connect

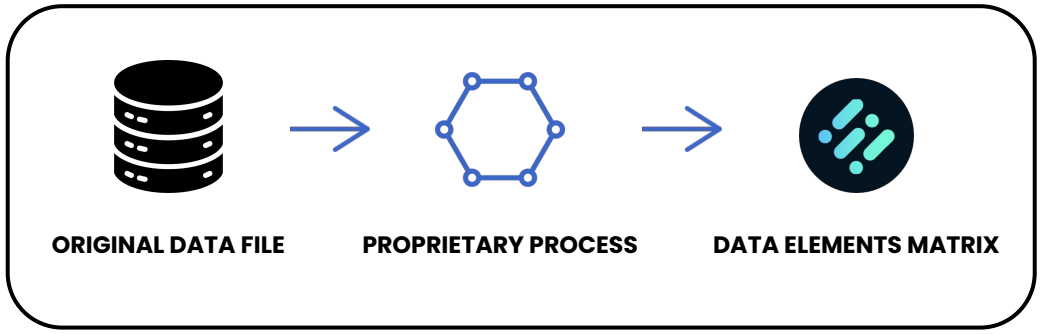
Data Collaboration & AI,

Without Data Movement

- Connects registry data globally - ***without moving the data***
- Standardizes data for greater Interoperability
  - common ontologies - SNOMED CT, LOINC, UMLS, etc.
- Offers real-time, privacy-preserving AI/ML analysis.
- Delivers on-demand, patient insights & RWE.
- Secure and compliant - HIPAA & GDPR (3rd party audited)
- Lightweight, scalable software—integrates seamlessly with existing IT systems.
- Compatible with all major cloud providers - Google, Amazon, Microsoft & Oracle.

Our Breakthrough Technology - ***The Data Elements Matrix (DEM)***

The Decentra Health team Invented a proprietary data mining process that converts patient data to a new privacy-preserving format - The DEM.



DEM is GDPR & HIPAA Compliant - Third party audited.

Avoids need to redo GDPR/HIPAA process for each study, as DEM Is compliant and therefore all output Is compliant by nature.

Unique Capabilities of The DEM

- The DEM is a true/full representation of your data
- Novel, built-in privacy preservation & anonymization
- Secure, global data connectivity & sharing
- Powerful AI/ML-driven analysis
- Auditable and traceable
  - All sharing & analysis is tracked & logged.

How Our Federated System Works

Connect registries globally without moving or exposing patient data.

Step 1

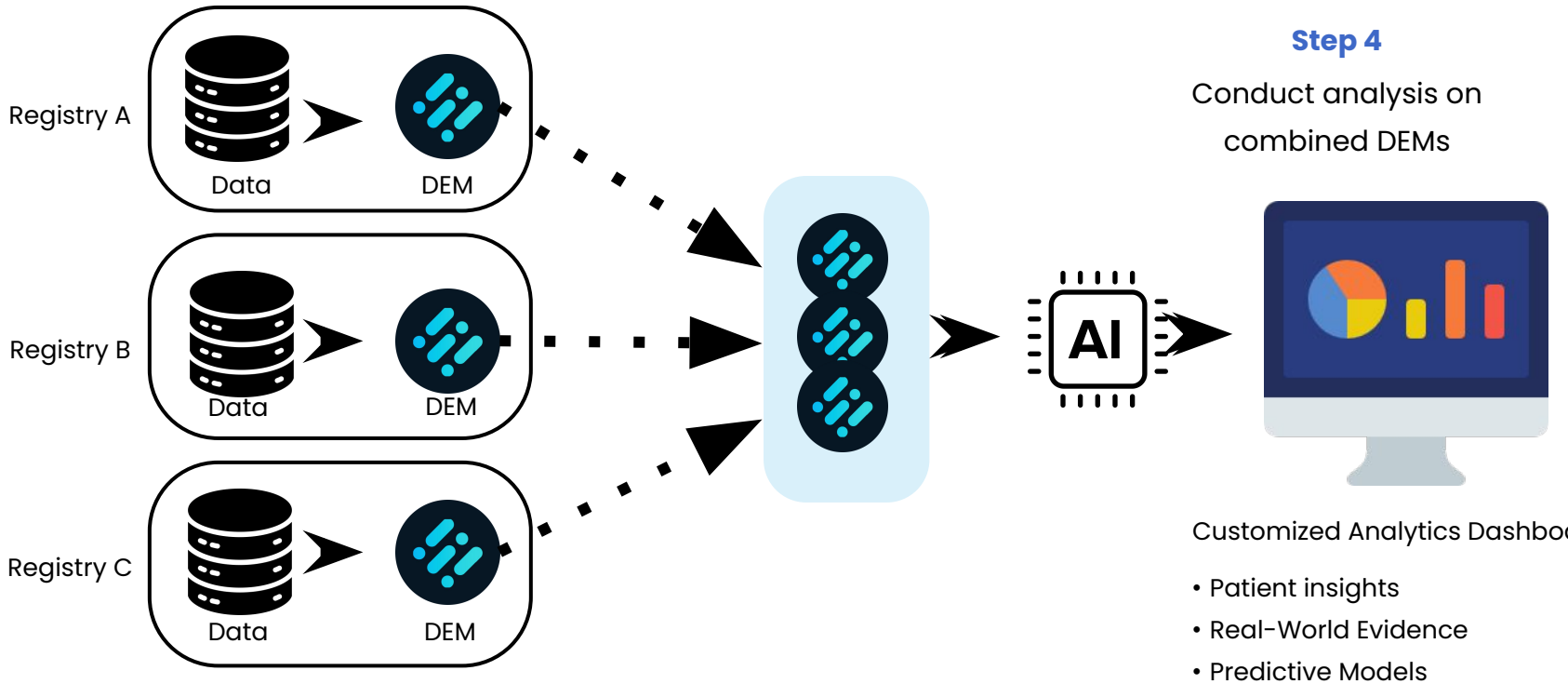
Install simple Decentra plugin at your local registry

Step 2

Harmonize data across all registries

Step 3

Our Decentra plugin automatically converts data to DEM format and connects DEMs across partner registries



Step 4

Conduct analysis on combined DEMs

Customized Analytics Dashboards

- Patient insights
- Real-World Evidence
- Predictive Models

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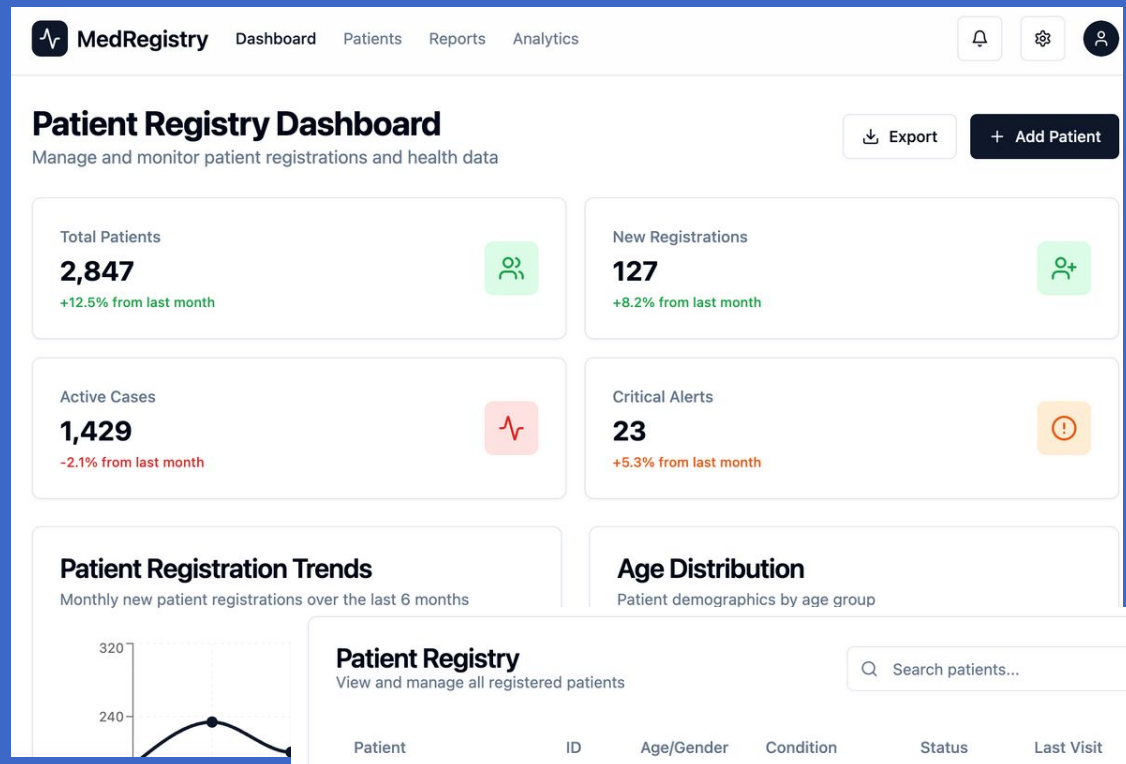
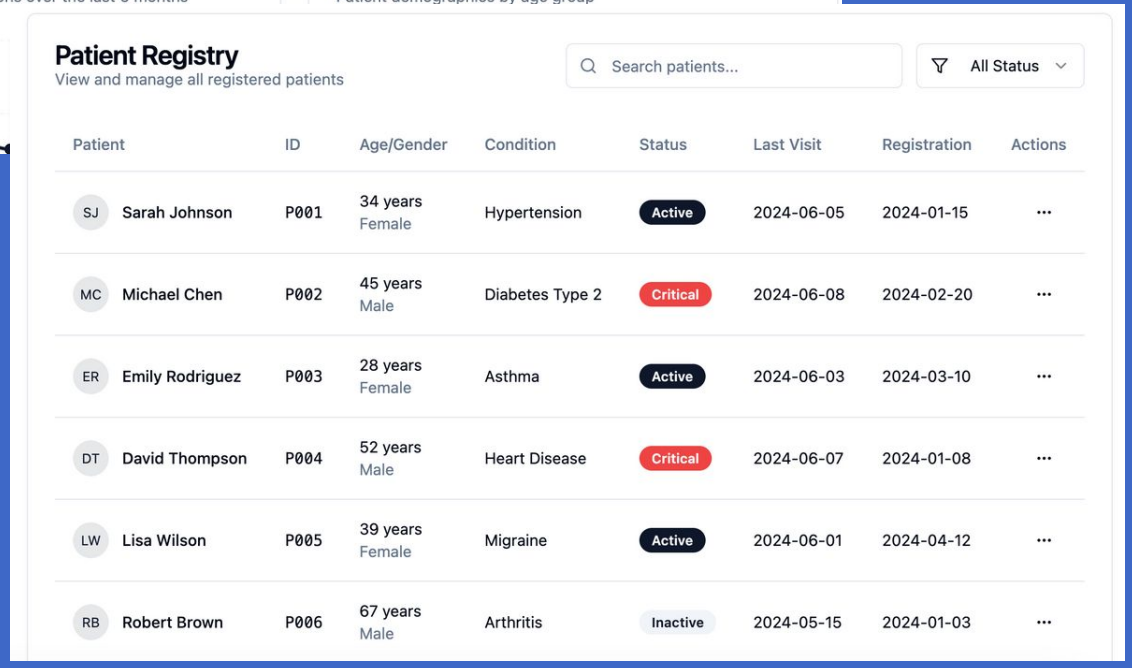
Registry

Decentra Health Is building a registry for FraXl that will be hosted In Belgium.

- **Flexible, Condition-Specific Design:** Customizable modules support clinician input, PROs, and direct patient data capture.
- **Real-Time Insights & Reporting:** Interactive dashboards provide instant access to trends, outcomes, and study metrics.
- **Secure & Compliant Infrastructure:** Built with privacy at the core—HIPAA/GDPR compliant, with granular access and

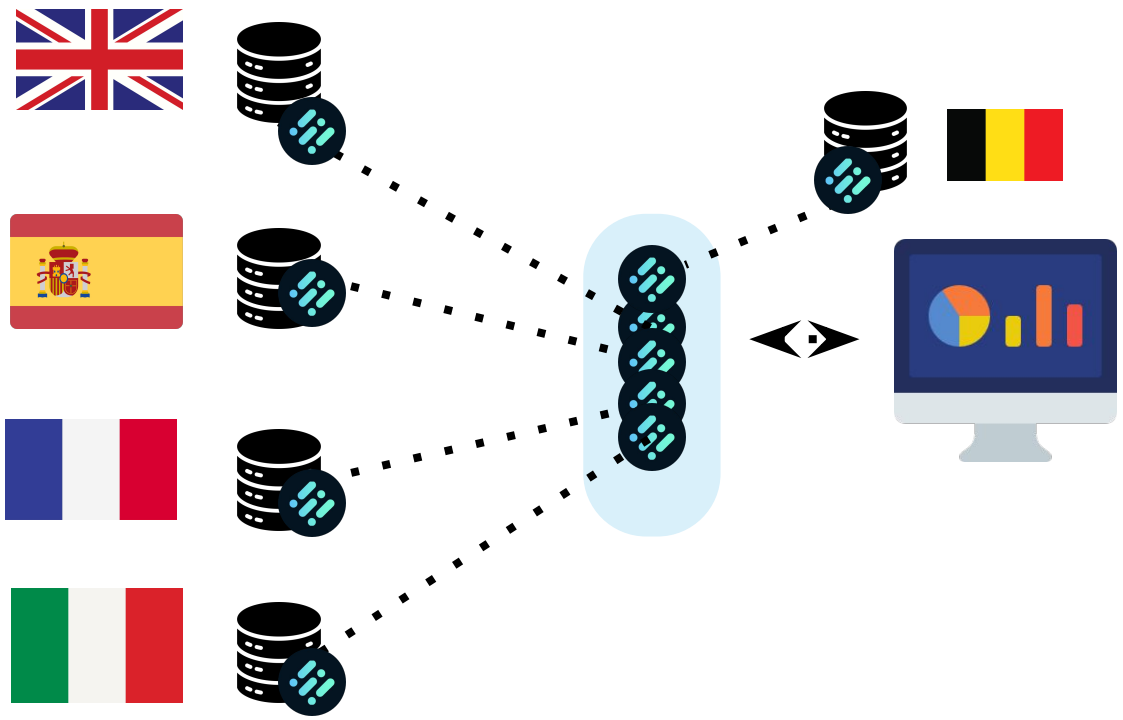




*Example Visuals

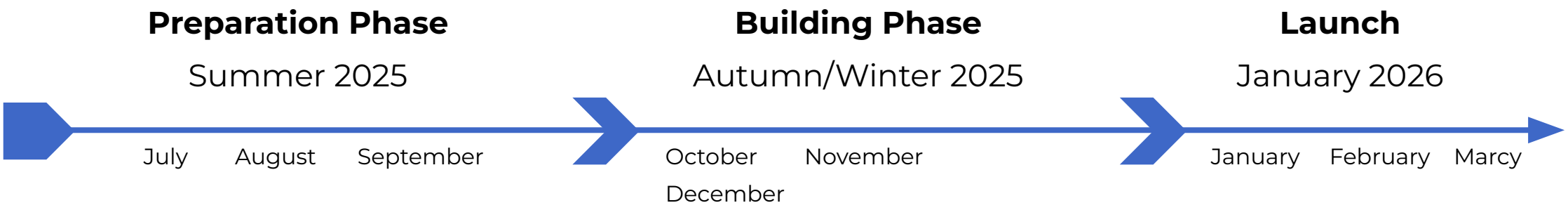
Fragile X Global Federated Registry Platform

Unlock collaborative research and move science forward.



FRAGILE X INTERNATIONAL

PROJECT TIMELINE



Preparation Phase

- Introduce plan to stakeholders
- FraXl setting up governance committee for oversight
- Education about technology & process

Building Phase

- Building infrastructure for federated registry
- Agreeing data points for FX registry
- Agreeing initial registries to join the fedrated system
- Testing & Initial pilots

Launch

- Launch FX federated registry platform

Thank You

Scan for Decentra Health's full slide deck.

Contact:

Kirsten Johnson
kjohnson@fraxi.org

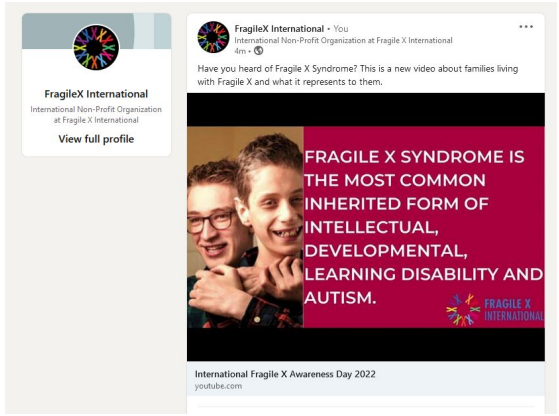
Social Media

@FragileXInternational

@fragilexint

@fragilexinternational

Fragile X International




Fragile X International
International Fragile X Awareness Day
2024
We are a force!
2024
Stronger than fragile x
Authentic stories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RESERVAT
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