



From Diagnosis and Research Readiness: A Parent-Led Implementation Model for Primrose Syndrome

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Background

Primrose syndrome is an ultra-rare, multisystem neurodevelopmental disorder caused by pathogenic variants on the gene *ZBTB20* (located at 3q13.31). Features include developmental, intellectual, and behavioral disabilities.

The severity and combination of symptoms vary widely. Families report isolation, fragmented care, limited access to knowledgeable clinicians, and few coordinated opportunities to participate in research.

Methods

This is a retrospective descriptive implementation assessment of Beyond Primrose ZBTB20 Foundation activities from November 2025 through September 2026. Activities and completed or initiated milestones were mapped across five domains: community engagement, governance, education and support, data infrastructure, and translational research.

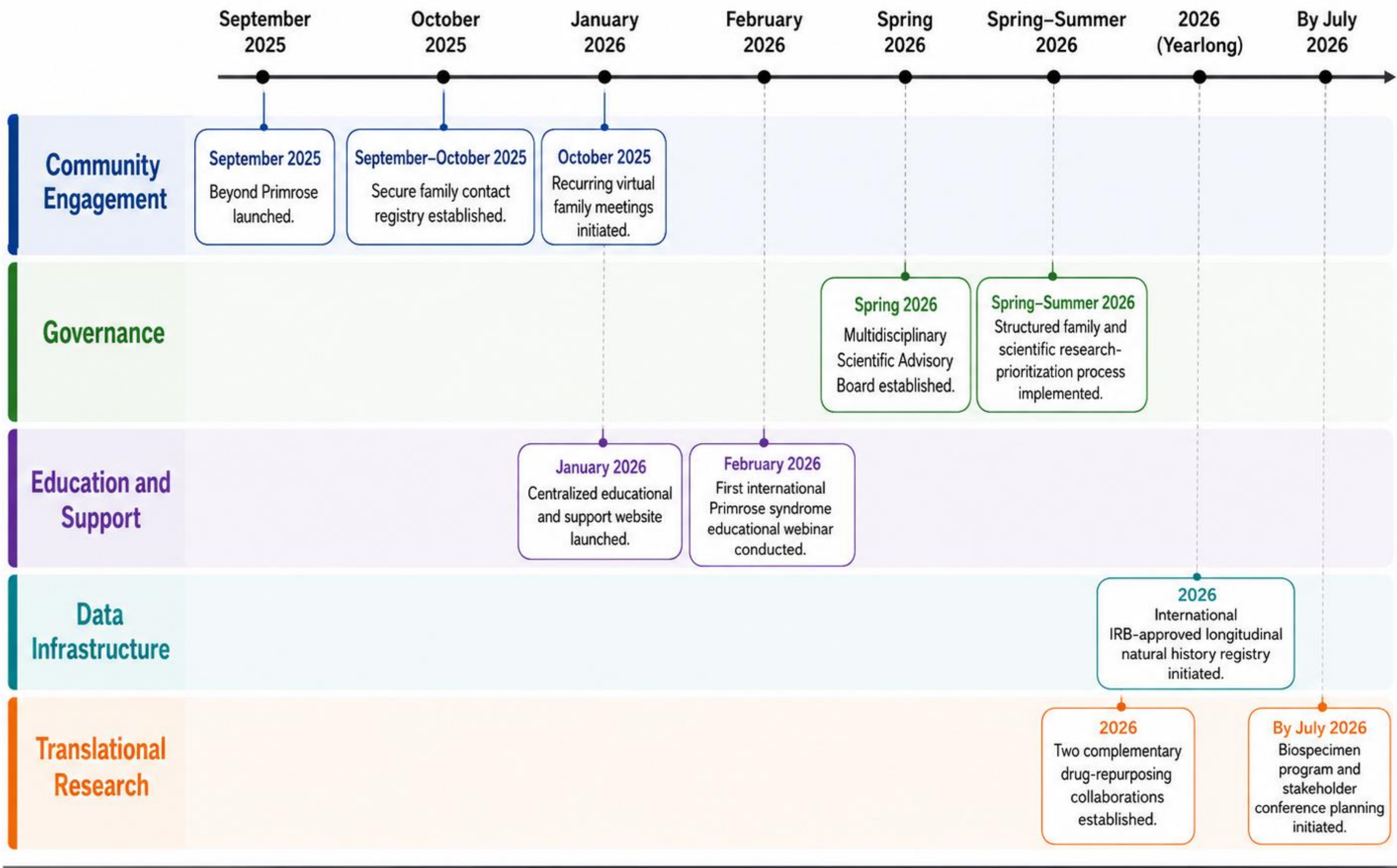
Results

Within 9 months, the initiative connected 88 affected families worldwide through a secure contact registry and established recurring virtual meetings. More recently, a communication platform "beyond primrose research and support network" has been implemented to allow families direct support and share experiences to better guide patient care. Research-readiness milestones included initiating an international, institutional review board-approved longitudinal natural history registry incorporating standardized measures.

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Figure 1. From Diagnosis to Research Readiness: Ten-Month Implementation Timeline, September 2025–July 2026



Within ten months, community engagement, multidisciplinary governance, education, data infrastructure, and translational research activities were developed in parallel, creating coordinated pathways from family connection to therapeutic research.

Table 1. Research-Readiness Outputs Achieved or Initiated During the Ten-Month Assessment Period.

Domain	Data source	Key output	Quantitative indicator/status
 Community Engagement	Contact-registry and meeting records	Secure global contact network and recurring family meetings	>100 affected families connected worldwide
 Governance	Advisory-board and meeting records	Multidisciplinary Scientific Advisory Board and joint priority-setting process	Established and operational
 Education and Support	Webinar and website records	International educational webinar, centralized website, caregiver resources	Webinar delivered; website launched; additional resources in development
 Data Infrastructure	Registry and collaboration records	International IRB-approved longitudinal natural history registry	Initiated
 Translational Research	Research milestone tracker	Patient-derived fibroblast and RNA-based drug-repurposing collaborations	Two complementary collaborations established
 Future Infrastructure	Planning records	International biospecimen program and multistakeholder conference	Planning initiated

Outputs were summarized descriptively across five implementation domains; future infrastructure activities are listed separately because they were initiated during the assessment period but are not part of the original five-domain framework.

Conclusion

A patient-led organization can build measurable research-readiness infrastructure on a compressed timeline when community engagement, multidisciplinary governance, education, data systems, and translational partnerships are developed in parallel. Consolidation of literature and data, along with lived experience, allows family utilization when communicating with providers is essential to producing therapeutically favorable outcomes globally.

