



TWO DECADES OF RESEARCH PROGRESSION ON LOEYS-DIETZ SYNDROME

**A PRODUCT OF LDS CANADA'S
GLOBAL RESEARCH AND INSIGHTS PROGRAM (GRIP)**

Background

Loeys-Dietz syndrome (LDS) is a rare, autosomal dominant connective tissue disorder caused by mutations in the TGF- β signaling pathway, leading to aggressive, multisystem vascular disease.

Since its discovery in 2005, research has progressed from gene identification and diagnostic criteria to advanced imaging, surgical optimization, and emerging translational therapies. This infographic synthesizes 1,275 studies (2005–2025), showing how these advances have transformed LDS into a manageable chronic condition, while revealing persistent gaps in quality of life and patient-centered outcomes.

SCOPE OF REVIEW



- 7,242 records identified
- 1,275 studies included
- 20 years of research

CLINICAL MANIFESTATIONS



- Cardiovascular
 - Aneurysms, dissections at smaller diameters
- Musculoskeletal
 - Hypermobility, scoliosis, chest wall deformities
- Neurologic
 - Headaches, neuropathic pain, dizziness
- Craniofacial
 - Hypertelorism, bifid uvula, cleft palate
- Reproductive
 - Pregnancy-related vascular risk
- Immunologic
 - Allergies, asthma, eosinophilic GI disease

WHERE THE RESEARCH FOCUSED



In the 1,275 studies reviewed, 6 main research themes were identified:

- Genetics & Mechanisms — 352
- Clinical Manifestations — 406
- Treatment & Management — 323
- Diagnostics & Imaging — 139
- Science & Technology — 38
- Patient Outcomes — 16



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DIAGNOSTICS & IMAGING



Before:

- Physical traits & clinical suspicion

Now:

- ✓ Genetic confirmation
- ✓ Whole-body MRI
- ✓ Mutation-specific management
- ✓ Earlier surgical thresholds (4.0–4.5 cm)

TREATMENT IMPACT



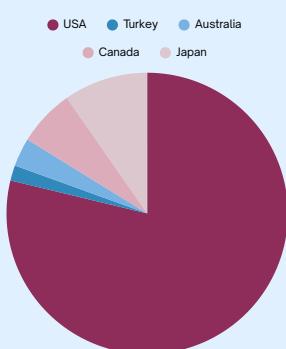
🔧 296 surgical studies

Key points:

- Optimized timing & techniques
- Improved perioperative outcomes
- Early pediatric intervention

✓ **LDS has shifted to a chronic,
 manageable condition**

LDS RESEARCH PUBLICATIONS IN GRIP PER GEOGRAPHY (TOP 10)



GENETICS → TRANSLATION



Research is moving from the lab to the patient:

Mutation discovery

↓
 Genetic testing & inheritance mapping

↓
 Mouse models

↓
 Early gene & drug therapy exploration

↗ **Toward preventive,
 molecular intervention**

THE GAP



What should research look into next?

🧠 Mental health

↗ Quality of life

👤 🚫 Long-term functioning

❤️ 💊 **Survival has improved —
 lived experience lags behind.**



The next era of LDS research must pair life-saving science with quality-of-life outcomes.