

Key Facts & Policy Recommendations



Rare Disease Overview

- There are more than **10,000 rare diseases**, affecting 25 to 30 million Americans—that's **1 in 10 Americans**. Many rare diseases affect the skeletal system.
- **80% of rare diseases are genetic**, and about **70% begin in childhood**. Approximately **two-thirds of Americans living with rare diseases are children**.
- Of the 10,000 known rare diseases, **95% have no treatment**. Patients are often treated “off-label,” leading to **insurance reimbursement challenges**.



Skeletal Conditions & Rare Disease

- Many **skeletal dysplasias and musculoskeletal disorders** fall under the rare disease umbrella.
- These conditions often require **multidisciplinary care**, specialized equipment, and **ongoing physical therapy**, which may not be readily accessible or affordable.
- A “**wait and see**” approach is too often applied, delaying diagnosis and access to interventions that could improve quality of life and long-term outcomes.



State Policy Priorities

- **Preserve disability programs and services:** Maintaining access to effective, existing programs provides a lifeline to California’s children and adults facing mobility challenges.
- **Provide support across key life stages:** Transitions through stages of life from home to school, pediatric to adult care, and school aged to employed, require accessibility modifications and other support services.
- **Sustain Medi-Cal access for children with physical disabilities:** Preserving access to necessary specialists, innovative treatments, home-based care, and adaptive devices is critical.



Bills to Support

- **Enable early and accurate diagnosis** with newborn screening and genetic testing to help parents understand and prepare for the progression of their newborn’s rare condition.
 - AB 242 (Boerner): Genetic disease screening
- **Support access to healthcare and mobility devices** amid ongoing, life-long medical need with limited treatment options to improve quality of life.
 - AB 539 (Schiavo): Health care coverage: prior authorizations
 - AB 517 (Krell): Medi-Cal: complex rehabilitation technology: wheelchairs