9 SIGNS OF A RARE DISEASE

WHEN TO LEARN MORE AND CONTACT A DOCTOR

Two or more of these signs may warrant a referral to your geneticist or to the UDN who are experienced in identifying rare diseases.

1. PHYSICAL FEATURES

Distinctive facial features, skeletal differences, or organ malformations.

2. DEVELOPMENTAL DELAYS

Delays in achieving or sudden loss of developmental milestones (speech, motor, or social).

3. INTELLECTUAL DISABILITY

Cognitive impairment or learning difficulties.

4. GROWTH CONCERNS

Unexpected inability to grow or excessive growth.

5. RECURRENT HEALTH ISSUES

Frequent infections, autoimmune problems, or other health issues.

6. MULTIPLE SPECIALIST VISITS

Visits to multiple specialists result in no diagnoses or variable, seemingly unrelated diagnoses.

7. FAMILY HISTORY

A history of unexplained health problems, birth defects, or developmental delays.

8. UNEXPLAINED SYMPTOMS

The presence of symptoms that cannot be explained by known conditions.

9. SENSORY CHALLENGES

Hearing loss, vision problems, or other sensory issues.

In the United States, a rare disease is as a disorder or condition that impacts fewer than 200,000 people across the country. There are more than 10,000 known rare diseases that affect about 1 in 10 people (or 30 million people).

PARTICIPANT ENGAGEMENT AND EMPOWERMENT RESOURCE

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