

Supplementary Table 2. Categories used to describe disease type of each proband

<b>Category</b>	<b>Description</b>
Allergies and disorders of the immune system	NA
Cardiology and vascular conditions	heart, artery, vein, and lymph disorders
Dentistry and craniofacial	bones of head and face
Dermatology	skin diseases and disorders
Endocrinology	disorder of the endocrine glands and hormones
Gastroenterology	disorder of the stomach and intestines
Gynecology and reproductive medicine	NA
Hematology	blood diseases and disorders
Infectious diseases	NA
Musculoskeletal and orthopedics	structural and functional disorders of muscles, bones, and joints
Nephrology	kidney diseases and disorders
Neurology	disorders of the nervous system, including brain and spinal cord
Oncology	tumors and cancer
Ophthalmology	eye disorders and diseases
Psychiatry	NA
Pulmonology	lung disorders and diseases
Rheumatology	immune disorders of the joints, muscles, and ligaments
Urology	NA
Toxicology and Environmental Medicine	NA
N/A	No information is available
Multiple Congenital Anomalies	NA
Other	none of the above categories