Genetic Variation within CdLS

By Dr. Antonie Kline, CdLS-USA Medical Director

Life is full of variation. No two people are completely alike, not even identical twins. This holds true in the field of genetics as well. With syndromes, there will always be degrees of involvement, differences in signs or symptoms, and a range of clinical effects. In CdLS, despite similarities, differences abound.

The source of variation is largely genetic. Many of the thousands of genes existing on our chromosomes do not carry out a major role in the body's functioning but act as assistants, influencing each job ever so slightly. Although we do not yet know where or how many genes there are that, when changed (mutated), produce CdLS, we do know they exist. Every individual with CdLS has his or her own panel of genes, inherited from both parents, that regularly influence the CdLS gene(s). Family background, ethnicity, and country of origin influence external appearance in everybody, including individuals with CdLS. Intelligence and level of developmental achievement can also be affected by family traits.

What strikes many families coming to a CdLS gathering for the first time is twofold: (1) individuals with CdLS largely resemble each other, particularly in their small size, facial features, extra body hair, and arm or hand differences, and (2) there is a broad difference in abilities at the same chronological age. What produces this difference is again unknown. Why are a small percentage of people with CdLS unable to walk? Why can a small percentage of people with CdLS attend a regular high school and college? We hope that answers to these questions will be discovered in the near future as molecular work progresses.

Individuals with CdLS who are said to be "mildly affected" or "less severely affected" are in the minority within our known group. Some joined the Foundation as infants when the features are easier to recognize. Others have joined in adulthood. Some of us believe that there are many people in our country (and likely the world) who have had only minor setbacks in life, either with medical issues or related to schooling, and who maybe are on the smaller side, who have CdLS but have never had it diagnosed. It will take the spreading of more awareness and the improvement of professionals' education to be able to recognize CdLS whenever it occurs.

Once the gene(s) is/are discovered, we will be able to verify the diagnosis of CdLS. The Foundation's mission includes helping people with the diagnosis of CdLS and others with similar characteristics make informed decisions throughout their lives and to ensure that support is provided to a maximum number of families needing intervention and resources. The web site should be increasingly helpful in this regard.