ARID1B Syndrome

https://pubmed.ncbi.nlm.nih.gov/30349098/

Purpose:

Pathogenic variants in ARID1B are one of the most frequent causes of intellectual disability (ID) as determined by large-scale exome sequencing studies. Most studies published thus far describe clinically diagnosed Coffin-Siris patients (ARID1B-CSS) and it is unclear whether these data are representative for patients identified through sequencing of unbiased ID cohorts (ARID1B-ID). We therefore sought to determine genotypic and phenotypic differences between ARID1B-ID and ARID1B-CSS. In parallel, we investigated the effect of different methods of phenotype reporting.

Methods:

Clinicians entered clinical data in an extensive web-based survey.

Results:

79 ARID1B-CSS and 64 ARID1B-ID patients were included. CSS-associated dysmorphic features, such as thick eyebrows, long eyelashes, thick alae nasi, long and/or broad philtrum, small nails and small or absent fifth distal phalanx and hypertrichosis, were observed significantly more often ($p < 0.001$) in ARID1B-CSS patients. No other significant differences were identified.

Conclusion:

There are only minor differences between ARID1B-ID and ARID1B-CSS patients. ARID1B-related disorders seem to consist of a spectrum, and patients should be managed similarly. We demonstrated that data collection methods without an explicit option to report the absence of a feature (such as most Human Phenotype Ontology-based methods) tended to underestimate gene-related features.