Autism spectrum disorders (ASDs) comprise an etiologically heterogeneous group of behaviorally diagnosed disorders defined by impairments in social skills, communication, and repetitive behaviors. Detection of biomarkers is a key tool in the search for biologically homogeneous ASD subgroups. Yao et al (2009) identified the pupillary light reflex (PLR) as a potential autism biomarker: A group of 22 children with ASD exhibited prolonged PLR latency and decreased constriction amplitude compared to 43 typically developing controls. PLR latency discriminated the ASD group from controls with a cross-validated success rate of 89.6%, which increased to 92.5% when constriction amplitude was considered. The ASD group also displayed significance variance in PLR parameters, which may indicate heterogeneity.

The purpose of this study was to identify clinical and etiologic variables in the children with ASD that correlate the variance in PLR parameters. Phenotypic features studied include dysmorphology, head size, growth, gender, family history of autism and related disorders, parental age at conception, age & type of onset, language development, improvement with fever, and neurologic history. Two neurologic features correlated with PLR parameters: Toe walking during development correlated with prolonged latency (p=0.01) and clinical improvement during febrile illnesses correlated with increased constriction amplitude (p=0.006).

These results raise the possibility of cerebellar and autonomic mechanisms underlying some ASDs. This study may guide future research concerning PLR as a possible biomarker that can identify homogeneous ASD subgroups and permit the discovery of more effective subgroup specific treatments and preventative strategies.