In two syndromic autism families, Ghasemi et al. discovered variants in two muscular dystrophy genes, DMD and CHKB. Given that DMD and CHKB are recognized for their participation in the non-cognitive manifestations of muscular dystrophy, it indicates that some genes transcend the boundary of apparently unrelated clinical categories, thereby establishing a novel connection between ASD and muscular dystrophy. The findings also shed light on the complex inheritance patterns observed in Iranian consanguineous simplex families and emphasize the connection between autism spectrum disorder and muscular dystrophy. This underscores a likely genetic convergence between neurodevelopmental and neuromuscular disorders [1)].

1)
Ghasemi MR, Sadeghi H, Hashemi-Gorji F, Mirfakhraie R, Gupta V, Ben-Mahmoud A, Bagheri S, Razjouyan K, Salehpour S, Tonekaboni SH, Dianatpour M, Omrani D, Jang MH, Layman LC, Miryounesi M, Kim HG. Exome sequencing reveals neurodevelopmental genes in simplex consanguineous Iranian families with syndromic autism. BMC Med Genomics. 2024 Aug 5;17(1):196. doi: 10.1186/s12920-024-01969-6. PMID: 39103847.