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Congenital ptosis, scoliosis and malignant hyperthermia susceptibility in siblings with recessive *RYR1* mutations

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The study aims to identify those children with ptosis and who are the candidates for ophthalmic surgery; who might harbor *RYR1* mutations, because intraoperative malignant hyperthermia is potentially lethal. We report for the first time congenital ptosis as the only ophthalmic findings in 2 siblings with scoliosis and underlying recessive *RYR1* mutations. Malignant hyperthermia susceptibility is a rare pharmacogenic disorder of skeletal muscle calcium regulation caused by mutations in the skeletal muscle ryanodine receptor 1 (*RYR1*) gene. It is important to identify those children who are candidates for ophthalmic surgery, who might harbor *RYR1* mutations.

Biography

Amani Al Bakri has completed her Ophthalmology Residency training in King Khaled Eye Specialist Hospital, Saudi Arabia. Currently, she is in her final year of Fellowship in Pediatric Ophthalmology and Strabismus, published few papers and is looking forward to publish more in ocular genetic field.

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