Abstract for UM symposium

TITLE: Case Report: CATCH-22 

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PURPOSE: To report a rare paediatric genetic disorder and its ophthalmic features.

METHOD: This is a case of a neonate who was referred to the ophthalmology department for bilateral proptosis. He had a dysmorphic facies and multiple limb malformations. Examination revealed complex multisystem involvement. Ophthalmological assessment was performed revealed the presence of multiple ocular abnormalities.

RESULTS: Ocular features seen in this patient include bilateral proptosis with shallow orbits, small partially formed eyebrows, mild lagophthalmos with mild exposure keratopathy, V-pattern exotropia and bilateral palish discs. Chromosomal study was normal with 46XY. FISH test (karyotyping) was consistent with with microdeletion of genes on chromosome 22 which confirms DiGeorge Syndrome.

CONCLUSION: We emphasize the importance of genetic studies to aid in the management of multisystem disorders. The role of the ophthalmologist would be in the recognition of abnormal ophthalmic features and manage them appropriately.

Keywords: Proptosis, DiGeorge syndrome