Format: Abstract

Biochem Biophys Res Commun. 2016 Aug 20. pii: S0006-291X(16)31282-7. doi: 10.1016/j.bbrc.2016.08.027. [Epub ahead of print]

Gallus gallus orthologous to human alpha-dystroglycanopathies candidate genes: Gene expression and characterization during chicken embryogenesis.

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Abstract

Alpha-dystroglycanopathies are a heterogenic group of human rare diseases that have in common defects of α-dystroglycan o-glycosylation. These congenital disorders share common features as muscular dystrophy, malformations on central nervous system and more rarely altered ocular development, as well as mutations on a set of candidate genes involved on those syndromes. Severity of the syndromes is variable, appearing Walker-Warburg as the most severe where mutations at protein o-mannosyl transferases POMT1 and POMT2 genes are frequently described. When studying the lack of MmPomt1 in mouse embryonic development, as a murine model of Walker-Warburg syndrome, MmPomt1 null phenotype was lethal because Reitchert's membrane fails during embryonic development. Here we report gene expression from Gallus gallus orthologous genes to human candidates on alpha-dystroglycanopathies POMT1, POMT2, POMGnT1, FKTN, FKRP and LARGE, making special emphasis in expression and localization of GgPomt1. Results obtained by quantitative RT-PCR, western-blot and immunochemistry revealed close gene expression patterns among human and chicken at key tissues affected during development when suffering an alpha-dystroglycanopathy, leading us to stand chicken as a useful animal model for molecular characterization of glycosyltransferases involved in the o-glycosylation of α-Dystroglycan and its role in embryonic development.

KEYWORDS: Chicken; Dystroglycanopathies; POMT1; Walker-Warburg syndrome

PMID: 27553274 DOI: 10.1016/j.bbrc.2016.08.027

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