

POSTER PRESENTATION

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# Using the *talpid*<sup>2</sup> as novel model for determining the cellular and molecular etiology of Oral-facial-digital syndrome

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## Objective

Oral-facial-digital syndrome (OFD) is a ciliopathy characterized by craniofacial abnormalities including cleft lip/palate, glossal defects, and absent/dysmorphic or supernumerary teeth. In addition, these patients have several other abnormalities typical of a ciliopathy including polysyndactyly, hypoplasia of the cerebellar vermis (molar tooth sign), cardiac defects and polycystic kidneys. Recently a subset of OFD cases have been linked to mutations in the centriolar protein, calcium C2-dependent domain containing 3 (C2CD3). Interestingly, our previous work identified a mutation in C2CD3 as the causal genetic lesion for the avian *talpid*<sup>2</sup> mutant. Based on this common genetic etiology, we re-examined the *talpid*<sup>2</sup> mutant for OFD-like phenotypes. We found that almost all phenotypes are conserved between *talpid*<sup>2</sup> embryos and OFD patients. In light of this finding we utilized the *talpid*<sup>2</sup> to examine the cellular basis for the craniofacial phenotypes present in OFD.

## Methods

Using both *in vivo* and *in vitro* methods we analyzed specification, migration, proliferation and differentiation of cranial neural crest cells (CNCC) when C2CD3-dependent ciliogenesis was impacted.

## Results

Our studies suggest that whereas disruptions of C2CD3-dependent ciliogenesis did not affect CNCC specification or proliferation, it did affect CNCC migration and differentiation. Migrating *talpid*<sup>2</sup> CNCCs were more disperse than control CNCCs and their migration was impaired.

Furthermore, *talpid*<sup>2</sup> CNCC derived cartilages are larger relative to controls.

## Conclusions:

Taken together, these findings suggest that the avian *talpid*<sup>2</sup> mutant is a bona fide, novel model for OFD and that aberrant CNCC migration and differentiation could contribute to the pathology of C2CD3-dependent human OFD.

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