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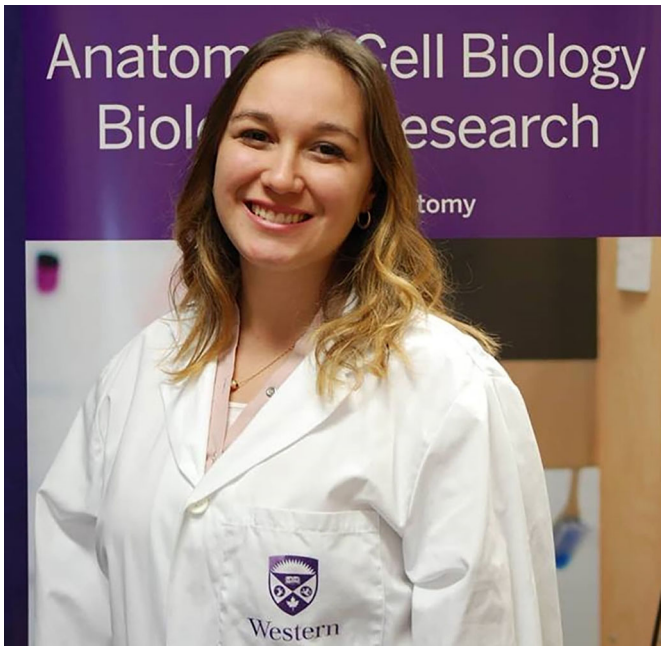
FIRST PERSON

First person – Julia Abitbol

First Person is a series of interviews with the first authors of a selection of papers published in Journal of Cell Science, helping early-career researchers promote themselves alongside their papers. Julia Abitbol is the first author on 'Mice harbouring an oculodentodigital dysplasia-linked Cx43 G60S mutation have severe hearing loss', published in Journal of Cell Science. Julia is a PhD student in the lab of Dale Laird at the University of Western Ontario, Canada, investigating the mechanisms of connexin-induced hearing loss.



Julia Abitbol

How would you explain the main findings of your paper to non-scientific family and friends?

Gap junction proteins allow our cells to communicate with one another, and are important for the development and function of many organ systems, including our ability to hear sound. We have known for many years now that mutations in certain genes encoding these gap junction proteins (specifically Cx26 and Cx30) cause inherited hearing loss. Our paper is the first to identify a mutation of a different gap junction family member, Cx43, that causes severe hearing loss. Furthermore, we were able to show that a Cx43 mutation will only result in hearing loss when the Cx43 functional status drops down to 20% of its normal function, implying that hearing is not only dependent on this gap junction channel but also on its functional status. When a child is born with congenital hearing loss, it is currently standard operating procedure to screen them for mutations in the gap junction family members that are known to cause hearing loss. Currently, we do not know the cause of congenital hearing loss in ∼25% of patients and, thus, this study suggests that there might be value in extending the screening panel to include Cx43, and in doing so possibly decrease the number of patients with congenital hearing loss due to unknown causes.

> **"Together, we've created excellent tools to be able to assess our research objectives in the best way we possibly can."**

Were there any specific challenges associated with this project? If so, how did you overcome them?

One of the biggest challenges I've faced so far is establishing a new niche in our lab that encompasses both cellular biology and the auditory system, and we are one of the few to do so in Canada. In forming this niche, I've learned and implemented a lot of different very intricate techniques, including culturing the epithelium of the postnatal cochlea to view our sensory hearing receptors, the hair cells (see picture below). Although there have been many challenges and hurdles along the way, we're in a great place to combine our lab's expertise on gap junction proteins with our collaborator Dr Brian Allman, who is an expert in the auditory field. Troubleshooting goes without saying in graduate studies, and if everything was easy, life would be pretty boring. Together, we've created excellent tools to be able to assess our research objectives in the best way we possibly can.

When doing the research, did you have a particular result or 'eureka' moment that has stuck with you?

For this project my 'eureka' moment was when the $Cx43^{G60S/+}$ mutant mice demonstrated such severe hearing loss. This was exceptionally exciting, not only because it was the first mouse model that I've tested to show hearing loss but also because it is the first documented Cx43 mutant mouse in which a substantial lack of Cx43 function causes severe hearing loss. From then on, we decided to pursue this study further, complementing it with another Cx43 mutant mouse that had a milder reduction in Cx43 function and that did not have hearing loss. Putting these models together, we were able to decipher that Cx43 was essential for hearing, but mice could tolerate a substantial reduction in Cx43 function before manifesting hearing loss.

Why did you choose Journal of Cell Science for your paper?

We chose Journal of Cell Science for our paper so that the general cell biology community would gain insight from these scientific discoveries. The fact remains that the molecular and cellular mechanisms of hearing loss are of fundamental importance and are not well understood. Mutations in the genes encoding gap junctions are the most prevalent cause of inherited hearing loss, with over 200 million carriers of these mutations worldwide. Because of this high prevalence, we believe it is of the utmost importance to describe these novel findings and target them to a cell biology readership, of which Journal of Cell Science is a renowned journal in the field. It is

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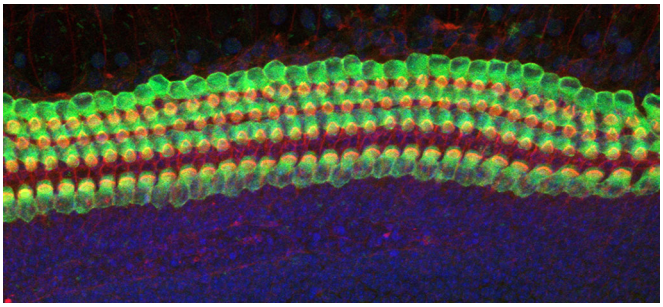

Postnatal culture of the sensory epithelium with hearing sensory receptors (hair cells) stained in green. Three rows of outer and one row of inner hair cells are visible

our hope that with increased knowledge and understanding of the basic cellular mechanisms underlying these mutations in hearing loss, we can start to intervene with therapeutics to treat hearing loss.

Have you had any significant mentors who have helped you beyond supervision in the lab? How was their guidance special?

I've been extremely fortunate to have a great team of mentors and lab personnel alongside me. My direct supervisor Dr Dale Laird and our collaborator Dr Brian Allman have contributed continual guidance throughout my studies and have led me to be the best researcher and critical thinker that I can be. In addition to supervisory roles, I've had the opportunity to be mentored by an extremely talented former postdoctoral fellow in my lab, Dr John Kelly, who trained me when I first started my graduate career. He has been instrumental in my success as a graduate student and taught me many of the intricate techniques relating to the auditory field that I've been able to implement in the lab. This mentorship was special in the sense that a challenging part of my graduate program thus far has been entering and setting up a new niche in our lab's field of study and he played a very important role in helping me do so.

"I'm excited to continue the scientific journey and discoveries ahead."

What's next for you?

I absolutely love science and could not imagine doing anything else. In what capacity I haven't definitively decided, but I'm excited to continue the scientific journey and discoveries ahead.

Tell us something interesting about yourself that wouldn't be on your CV

Back when I was at home in Sudbury I took many different types of dance classes and even acted as a teaching assistant in my later years there. Since I've been away from home for the past 8 years now I've turned my energies to intramural sports and going to the gym.

Reference

Abitbol, J. M., Kelly, J. J., Barr, K. J., Allman, B. L. and Laird, D. W. (2018). Mice harbouring an oculodentodigital dysplasia-linked Cx43 G60S mutation have severe hearing loss. *J. Cell Sci.* **131**, jcs214635.