

RYRI: A story about calcium channels and the people who have them

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RYRI encodes the ryanodine receptor – a calcium channel found in skeletal muscles which opens in response to sarcolemma depolarization allowing calcium to move from the sarcoplasmic reticulum into the cytoplasm. Those with *RYRI* mutations experience central core disease, a congenital myopathy which is characterized by profound muscular flaccidity. To a young medical student this phenomena is ‘interesting’, a label reserved for the few and far between cases in medicine when a clear cellular mechanism produces intuitive effects phenotypically. However, no amount of intellectualization can prepare medical students for the sight of a four-year old girl on an intubator who sits completely still in her room- the only movement coming from her eyes as she tracks the new presence in her space. Students may find this sight so uncanny that they quickly scan the patient’s medications to check for paralytics having already forgotten what they’ve just looked up regarding the effects of *RYRI* mutations.

Students are encouraged to push this unease to the side and proceed with their duties, checking with the mother for overnight updates and performing a physical exam. Diligently collecting data points for the all-important presentation: one of the many metrics by which students are measured.

Speaking of that all-important evaluation, when rounding in the pediatric ICU, students may be surprised

to find that their attendings call the parents to listen in while presenting outside the patient’s room. Students may find the presence of a parent to be abnormal, and their presentations may suffer for it- punctuated by pauses that linger slightly too long. Pauses in which the movie playing in the patient’s room can be heard. She’s watching Moana.

“ *See the line where the sky meets the sea? It calls me
And no one knows how far it goes
If the wind in my sail on the sea stays behind me
One day I’ll know-* ”

But this isn’t the time for listening to songs or watching movies. There are attendings to impress, patients to round on, and maybe even lives to save. And reader, if this choice of movie seems inappropriate or cruel, worry not- according to Glenwright et al. in the *Journal of Child Language*, we do not develop a sense of irony until age 5-7. That means, at least for the next 12 months, your hospitalized patient with an *RYRI* mutation will be untethered by anchor of prognosis. She’ll watch Moana and think to herself, “when I go there’s just no telling how far I’ll go.”

References

Glenwright Melanie, Pexman Pennym. Development of children’s ability to distinguish sarcasm and verbal irony*. *Journal of Child Language*. 2010;37(2):429-451. doi:10.1017/S0305000909009520