Case study: Anophthalmia

Learning objectives:

1.1.1 Describe how the nervous system is vital to the maintenance of homeostasis.
1.2.1 Describe the anatomical organization of the nervous system into central nervous system (brain and spinal cord) and peripheral nervous system (cranial and spinal nerves).
1.2.2 Describe the functional organization of the nervous system into the central nervous system and peripheral nervous system (afferent and efferent divisions).
1.4.3 Locate and give the roles of each of the major parts of the forebrain, midbrain and hindbrain.
1.5.1 Describe the major sensory and motor pathways.

Consider reviewing the following topics:

Organization of the nervous system, white matter fibers, anatomy and functions of the forebrain, midbrain, and hindbrain, and the cortical areas (see lecture notes “Nervous system: brain” and the associated video lectures). Consider completing the “Organization of the brain” and “The brain” worksheets.

Introduction

Patient story:

“Your child has anophthalmia, a syndrome characterized by the absence of eye tissue development. As a result, your son is blind. There are several causes for this syndrome, a genetic link being the most probable. We can run some genetic tests if you’d like.”

“Yes, maybe. I don’t know. We’ll think about it, doctor,” replied Anika.

“Caring for a blind child does present some difficulties, but we have several resources that’ll make sure you’re amply supported. We might want to first consider aesthetically shaping his orbital sockets, so they develop as his skull grows. This’ll ensure that prosthetic eyes can be used later on before he starts school,” offered Dr. Verma.

“Hmmm, yeah. Is this common? I mean, is this how things normally progress?” asked Anika.

“Yes, of course. We’ll get you in touch with an ophthalmologist and a paediatrician. From what I’ve read, they’ll do an ultrasound to examine the orbit. This syndrome is commonly associated with cysts that may have some capacity for vision. We’ll know for sure by checking for functional photoreceptors with something called a flash visual evoked potential (VEP) test. Sometimes, anophthalmia is also accompanied with brain atypicalities, so we’ll run an fMRI to check for those. Then we need to pay close attention to the development of his facial features.”

“What? Why?”

“The eyes are important for the development of the skull. When the eyes aren’t present, the sockets, like the orbital bones, usually remain underdeveloped, and this also compromises the use of prosthetics later in life. We’ll likely start with the use of expanders, which should help the eye socket grow with the rest of the skull. We’ll change these every few weeks in the first two years of his… um, does he have a name?”

Anika and Kevin glance at each other momentarily and smile. “Alex.”

“That’s a nice name. We’ll have to change these expanders often during Alex’s first two years, and less often after that. We can then get fitted prosthetics before he starts school. When he’s much older, surgical adjustments can be made if he chooses, like in the case of drooping eyelids. Otherwise, we’ll monitor Alex

during his development. If you want, we can eventually talk about arranging an appointment with a medical geneticist."

Kevin exhaled and straightened his back. "Okay. This is good. We'll be good. What kind of resources should we consider applying for?"

"Great question. I think the ophthalmologist ought to know more than me. I'm from the UK, and there, you can typically apply for a Certificate of Visual Impairment. I'm sure there are equivalent documents here. You'll want this to expedite some of the services offered. It'll be important to learn how to interact with your child as he develops into early childhood. Consider that the majority of our learning in schools is visual, so special school education programs are a good bet." Dr. Verma clasped her hands together and smiled. "We have great resources for visually-impaired children."

Anika shifted in her chair, glanced at Kevin, and quickly cocked her head towards Dr. Verma. "You mentioned something about interacting with Alex as he grows up?"

"Yes. I don't know much about childhood brain development in visually impaired children, but I do know a little bit. You'll want to maintain a stream of language since this is going to be Alex's link to the environment. Describe the toys he's playing with, the objects he touches. Try to maintain a constant environment in the house. This ought to encourage exploration as Alex learns to crawl and walk. That's more or less what I understand. Your ophthalmologist will know a bit more."

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Anika, Kevin, and Alex visit Dr. Yazzie, the ophthalmologist, as Alex is about to enter middle school. "How are your studies, Alex?"

"Fine. Things are good. I'm enjoying science. I heard we get to learn about space this year." Alex shifted forward and gripped the edge of his seat. "You know, learn about black holes and pulsars, and stuff."

"Yeah, I was never all that interested in physics, to be honest. But biology! Anatomy! Medicine! Now, that got my attention. Wouldn't you prefer a job in medicine?" Dr. Yazzie slyly glanced at Anika and Kevin, who let out a small chuckle.

"Nope. That's boring. Not for me."

"Well, that's a shame. But I still have time to convince you, yet," ending with an overdramatized display of frustration.

"He can't be helped," chimed in Kevin.

"So! You've met with the medical geneticist? Did he identify the gene?"

Anika slid her shoulder bag onto the corner of her chair, and comfortably shifted in her chair. "Yes, she did. PAX2. P-A-X-number 2. She mentioned it was a transcription factor. Luckily, Alex hasn't shown any kidney defects, so it seems to have only affected eye development."

"Okay, that's good. I'm glad you went through with it. That brings us to today's little adventure. Do you understand how an fMRI works, Alex?"

"Kind of. It's like an EEG, right?"

Dr. Yazzie clicked her teeth and leaned back in her chair, tilting her head back as she stared at the ceiling. "It's interesting that you know about EEGs. They're not exactly the same, but they both measure brain activity. EEGs measure electrical activity while fMRIs record blood flow. An increase in blood flow in a particular area of the brain suggests increased activity in that area. Today, we'll be looking at your brain activity as you echolocate, along with some other activities, like listening to speech or music, and grasping objects."

"What do you think we'll find?" Kevin placed his arm around Alex's shoulder and ruffled his hair.

"To be honest, we're not sure. The research associate wants to see how Alex's cortical areas have changed on account of his blindness. Echolocation in people isn't common, so we'd like to know how sounds are used as Alex maps out his environment. For example, Alex uses sound to map out a room, whereas other people use visual information. That will certainly have an impact on the development of signals across Alex's

brain. The interesting thing about all this is that Alex doesn't have heightened auditory senses. As in, he hears as well as other kids his age, but we suspect he uses auditory signals a little differently. Do you have any questions, Alex?"

"Not now. Maybe after."

"Well, then. Let's go visit the staff and get you ready."

Questions to consider (will not be graded):

1. Why does Anika bring up Alex's kidneys when talking about the gene mutation?
2. Which of Alex's cortical areas are likely to exhibit atypical activity? Why?
3. Can you think of any other ways parents can help with the development of a visually impaired child?

References:

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