Eye & Ear Issues in Ichthyosis Patients

CME Series on Epidermal Differentiation Disorders — Presented by Dr. Lara Wine Lee

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Welcome to Eye & Ear Issues in Ichthyosis Patients, part of First's CME series on epidermal differentiation disorders or ichthyosis. Presenting this topic is Dr. Lara Wine Lee. Dr. Wine Lee is a professor of dermatology and pediatrics at the Medical University of South Carolina, where she has practiced since 2014. She cares for children with genetic skin disorders as part of the pediatric dermatology program.

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Well thank you so much for having me to discuss the topic of eye and ear issues in ichthyosis patients. I have disclosures related to clinical trials work in Leo Pharmaceuticals and Mayne Pharmaceuticals.

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Our goals today are to have a discussion about the eye and ear complications of patients with wide ranging EDDs, including both ichthyosis and palmoplantar keratodermas, and to have an understanding about the referral pathway and when a specialist would need to be involved in the care of these patients. So starting with the ear problems that we encounter in our patients with EDDs.

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So throughout different types of ichthyosis, the vast majority of patients do report ear problems including sensorineural hearing loss. And I think it's helpful to categorize the ear issues into three main categories. So one is external canal disease. And this has really to do directly with the problems that are seen within the skin where you can have obstruction of the ear canals and conductive hearing loss.

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There is inner ear disease, oftentimes directly related to the genetic basis of the EDD and then external ear abnormalities. And we'll go in detail about each of these different areas. So in terms of the external auditory ear canal problems, this actually occurs in greater than 80% of all patients with ichthyosis shown here in the diagraming. See the structure of the external ear canal.

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And what happens is just like there is scale production on the exterior skin, the auditory canal is a keratinizing surface as well. So there's shedding of the skin layers into the canal. And then this mixes with the wax and other debris that builds in our ear canal, and can lead to fairly severe impaction,

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which, if not removed or treated, can lead to these recurrent obstructive plugs.

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This is particularly accentuated in smaller children simply because of the small size of the ear canal, and then some types of ichthyosis as well that has some developmental abnormalities of the ear. The canals are even smaller. So what happens over time when you have these wax plugs that build up with the scale, is the patients can feel a lot of fullness, pruritus pain, and actually fairly severe obstructive hearing loss.

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Long term sequelae include recurrent infections that actually can even lead to scarring and further reduction in the size of the ear canal, and then recurrent ear infections, as well as the difficulty of diagnosing an external ear canal because of the obstruction and the internal, the otitis media, so an internal infection, oftentimes like in your office, you'll have problems with a diagnosis because the ear canal is obstructed.

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So just showing some pictures of what you may be dealing with. Here at the top you can see a keratinaceous plug in the external canal of a patient with autosomal recessive congenital ichthyosis. You can see that debris that really has obscured the whole canal. It actually can even push on this the size of the canal. And then once that plug is removed, there's a lot of inflammation and still a lot of scaling that remains.

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And this is this inflammation and the skin infection risk that you have anyway with ichthyotic skin can lead to infections of the ear canal that do require treatment as well. And then over time again some changes in the shape of the canal. It is important, therefore, to really expectedly discuss the management strategies to avoid this before it becomes a problem.

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So first of all, all visits should include an examination of the ears and when possible, an examination of the external auditory canal. And do this even when your patients aren't saying that there is a symptom. Particularly, our youngest patients may have difficulties expressing ear pain or ear fullness. So it's going to be important part to always consider in your visit.

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We do recommend debridement and that can be done in several ways. There are some families and patients that can develop techniques to do debridement and maintenance at home. But it's really, really important to counsel or involve your ear, nose and throat colleagues to talk about appropriate instrumentation of the external auditory canal. This is just one device that is a very safe one to use at home, but there are several options that you can find.

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And again, under the guidance of medical attention, sometimes specialists care. And do consider over time the point that you might actually need to look at hearing tests. So particularly in those times when children are acquiring language, any type of conductive or other hearing loss can really lead to problems with language development. Very regular use of gentle products that can reduce the wax buildup and reduce the plug can be helpful in between doctor's visits, or in between any home type of debridement techniques that they are using.

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And there are several examples of that with different brand names, but there are drops that you can buy over-the-counter. And then the potential use for liquid colace, particularly if they're not tolerating some of the other drops followed by irrigation once you soften up those plugs.

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The problem with the scaling and plugging of the canals can also be compounded by developmental changes in the ears that can be found in both syndromic and non syndromic forms of EDDs. And just some examples here across different types of EDDs where you have some, you know, frank, rudimentary development of the ears with the small canals, with the helix of the canal being abnormally formed.

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It is important to also note that if you're having difficulties with your clinical diagnosis, sometimes these specific ear malformations can help point you in a certain direction. But these are just a few examples and happens across many, many different types of EDDs. But it's also really important to remember that not all hearing loss in ichthyosis and palmoplantar keratodermas are due to actually obstructive hearing loss.

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There is a strong association with sensorineural hearing loss in several types as well. And the most prototypical type to think about here is going to be KID syndrome. The keratitis, ichthyosis and deafness where you have the combination of that triad there where they have very, very profound sensorineural hearing loss that is progressive. And the average age of onset of deafness is actually in those toddler years.

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So very important at the time to note that if you have the correct diagnosis and make sure we're getting appropriate care, because this is such a critical time for language development, which can then again be affected by your hearing problems. There is a number of types of EDDs that are related to the GJB2 mutation, and all of them have a variety of hearing loss that is possible.

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So getting the genetic diagnosis can really help you get ahead of that. And luckily we're in a time right now where there's lots of interventions that can be done with our audiology and our ENT colleagues so that these patients can maintain their hearing and not have that be such an issue over their lifetime. Important to note, in all of these forms that have different types of sensory neural hearing loss,

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we certainly don't want to compound the issue about conductive hearing loss with the scale buildup and the obstructive plugging. So very, very important to stay on top of all of that as well. So we don't have both types of hearing loss making the experience of the hearing so much worse for the patients over time.

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So moving on to the other very prominent system that is involved in many different types of patients with ichthyosis are going to be the eye involvement. And this very frequent as well. And it really is across all types. So with a lot of variability again, both syndromic and non-syndromic forms, the eyelid manifestations are one of the most important parts of the disease.

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And something that we really do have to expectantly manage again to prevent long term sequelae. So even with, you know, from less severe to more severe types, there's a lot of scaling that occurs on the eyelid and loss of the eyelashes over time as well, partially having to do with that inflammation that happens at the eyelid margin. And then ectropian which is that out turning of the eyes, exposing it to, again, the elements that kind of leads to that inflammation and scarring over time.

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So all of this leads to a lot of eye irritation. And oftentimes the severity of this may be actually linked to the severity of the skin manifestations of the type of ichthyosis. However, even in less of your types, you can see this to some extent. So a lot of variability here but very very important to preserve vision. So when you have the eyelid involvement over time you get the inability to completely close the eyes.

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So the picture here is a patient with ichthyosis. This is actually an attempt to completely close the eyes. And you can see that the eyelids don't actually oppose to protect the eye. So during sleep you really have to do protective eye lubricants and other types of stuff to prevent that, what we call exposure keratopathy. Because over time that will lead to inflammation, infection, scarring and even potentially perforation with the sequelae of being loss of vision, photophobia or other issues with the eyes.

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In addition, there are glands

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in the lower and upper eyelid that secrete the substance that keeps the homeostasis of the conjunctiva at an ideal level. Those are the meibomian glands, and in many types of ichthyosis these can have dysfunction as well. Some of it is just from plugging of those areas and then inability to kind of secrete the gland fluid.

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However, there's probably also some developmental changes that can happen related to the genetics of it as well. Similarly to the exposure keratopathy, you can get that photophobia erosions over time leading to scarring. Interestingly, the meibomian gland dysfunction leads to actually interning of the eyelid, which is entropian, but equally can contribute to eye problems as the ectropian that we had seen in the previous pictures.

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There also is really maybe some specificity to different types of syndromic ichthyosis as well, where the glands may be abnormally formed related to the genetics of it, predominantly seen in KID or photophobia and IFAP, which the P is photophobia is very central to the consequences of that type of ichthyosis.

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And it's always important to note some more specific types of eye findings that we can see that are specifically related to the genetics. And this is something to educate your patients on because when they do go see their eye doctors, many times eye doctors may not actually have many patients with different types of EDD And so it's important to understand which is related to the genetics of the disease, which may need management, and which could be not as consequential to the patients.

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So perfect example of that are going to be these corneal opacities and X-linked ichthyosis or STS-sEDD. Where they are not symptomatic, they do not affect vision, but they may be noticed by doctors and therefore might be concerned that there could be another cause of it. So it's important that throughout the crosses of types of ichthyosis, there's both eye issues related to the actual scaling and the shedding of the skin and ones that are going to be also genotype specific, furthering the importance of getting a correct genetic diagnosis to understand kind of counseling and management over time. In KIDs syndrome also, which again has a lot of eye manifestations, there is a progressive vascularizing keratopathy

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that leads to that photophobia irritation and reduced vision. So very very critical for these patients to continue regular eye follow up so that we're getting ahead of any issues that could threaten vision over time. So how does this interplay with the kind of progressive changes that we need to prevent to the eyes and the corneal abnormalities that if we're not getting ahead of some of the scarring and ectropian issues, the corneal opacities, ulceration and perforation can lead to scarring in up to 30% of patients.

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And again this is across severity. So while it is more likely if you have more severe skin manifestations of your ichthyosis, it can happen even in more mild patients. And very important to consider at what point your eye specialists do need to be involved to monitor. These are some more dramatic examples here where you can really see the scarring.

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You can see the corneal opacities and see the problems, understand the problems, how vision could be affected. But you really want to catch it before this steps. And that's where ophthalmologists are going to be very, very helpful to you. So I do recommend patients have regular follow up. The good news is that there are lots of interventions that can help us get ahead of the problems.

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So treating the scaling of the skin around the eyes can help with the inflammation and the ectropian and the loss of eyelashes. And remember eyelashes just protect our eyes from debris. So for instance, really liberal use of eye lubricants to protect the eyes, particularly if you're having the issues with the eyes closing fully. And then topical and systemic retinoids can help loosen up that skin,

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So we're not having that pulling on the eyelids. That leads to the problems over time. And this is a great example that was published showing how the use of actually topical tazarotene applied to the eyelid area can actually reverse some of those changes that we had seen, allowing more full closure of the eyes and protection then of the conjunctiva from development of scarring over time.

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In cases where we've not been able to prevent this, there are surgical correction options to return eyelid function to normal and restore vision capabilities.

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So when considering this all together, it's just important to remember that eye and ear disease are exceptionally common in our EDDs and that routine surveillance and involvement of your specialist really happen even prior to the development of any problems. There may be clinically significant disease even prior to any symptoms. And again, particularly in our young kids who may not have the words to appropriately tell us our symptoms, definitely consider this in your evaluation.

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The obstruction infection can lead to conductive hearing loss. This can be a large problem over time, even with compounding that with sensorineural hearing loss, and prevention of any of those eyelid abnormalities helps serve the vision over time. The phenotype, though, absolutely does matter, and getting an appropriate diagnosis can really help us know what the comprehensive care that is needed so that we can work with those that might have progressive sensory neural hearing loss or more of those eye abnormalities to kind of preventively and expectantly manage those.

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And then just make sure to have a good relationship with the ENT and the ophthalmology colleagues that work with it, so that we can take the best care of our patients as possible. Thank you very much.

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