

Kartagener Syndrome (Primary Ciliary Dyskinesia)

Kartagener Syndrome, also known as Primary Ciliary Dyskinesia, is a disease inherited in an autosomal recessive pattern. Patients with this disease have immotile cilia due to a Dynein arm defect. The defective cilia leads to decreased fertility due to immotile sperm and dysfunctional fallopian tube cilia. Patients with this disease also have an increased risk to suffer from ectopic pregnancies. The defective cilia can also lead to bronchiectasis, recurrent sinusitis, chronic ear infections and conductive hearing loss. Kartagener Syndrome is closely associated with the condition known as situs inversus. Testing for decreased nasal nitric oxide is used for screening.



PLAY PICMONIC

inheritance pattern

Autosomal Recessive

[Recessive-chocolate](#)

Kartagener Syndrome is a disease that is inherited in an autosomal recessive pattern. In an autosomal recessive disorder you inherit two mutated genes, one from each parent. These disorders are usually passed on by two carriers. With each pregnancy, two carriers have a 25 percent chance of having an unaffected child with two normal genes, a 50 percent chance of having an unaffected child who is also a carrier, and a 25 percent chance of having an affected child with two recessive genes.

pathogenesis

Immotile Cilia Due to a Dynein Arm Defect

[Roped-celery-guy by Dyed-nine with Falling-arm](#)

Dynein is a family of cytoskeletal motor proteins that move along microtubules in cells. They convert the chemical energy stored in ATP to mechanical work. Dynein transports various cellular cargos, provides forces and displacements important in mitosis, and drives the beat of eukaryotic cilia and flagella. All of these functions rely on dynein's ability to move towards the minus-end of the microtubules, known as retrograde transport. In Kartagener Syndrome, the cilia on the patient's cells are not motile due to the defective dynein arm.

Symptoms

Decreased Male and Female Fertility

[Down-arrow and Male-symbol and Female-symbol Fertility-flowers Withering](#)

Due to the dynein arm defect, in males with Kartagener Syndrome the cilia of sperm lose their mobility making it difficult for fertilization to occur, leading to decreased fertility. In females there is reduced fertility due to defective ovum transport because of the dyskinetic motion of oviductal cilia.

Immotile Sperm

[Roped-sperm](#)

In Kartagener Syndrome, the cilia that lines the flagellum of sperm cells is defective due to the dynein arm defect, which causes sperm to be immotile.

Dysfunctional Fallopian Tube Cilia

[Uterus holding a Snapped-celery with its Fallopian-tube](#)

Females with Kartagener Syndrome have a defective ovum transport because of dyskinetic motion of oviductal cilia caused by the dynein arm defect.

Increased Risk of Ectopic Pregnancy

[Up-arrow-risk Egg-top Pregnant-woman](#)

Ciliary motility plays a part in the transport of the ovum through the fallopian tube. The fertilized egg is supposed to then travel to the uterus 3 to 4 days after fertilization. Due to the ciliary defect in the fallopian tube in Kartagener Syndrome patients, the fertilized egg may stay in the fallopian tube, leading to an ectopic pregnancy.

Bronchiectasis

[Broccoli-with-tassels](#)

Cilia in airways help to clear mucus, dust and microbes out of the airways. In Kartagener Syndrome, the defective cilia can lead to infections that can damage the walls of the airways which leads to bronchiectasis.

Recurrent Sinusitis

[Sinner with Recurrent-clock](#)

In Kartagener Syndrome, the defective cilia can lead to infections due to them not being able to clear out microbes causing patients to develop sinusitis recurrently.

Chronic Ear Infections

[Old-crone with Ear alongside Bacteria-guys and Viruses](#)

Chronic ear infections can be seen in patients with Kartagener Syndrome due to the defective cilia in the ear. This prevents the effective clearance of the mucus and microbes from the ears which causes a higher chance of having chronic ear infections. Sinuses and the middle ear are connected, which means that sinusitis can lead to ear infections, which is also common in these patients.

Conductive Hearing Loss

[Musical-Conductor with Plugged-ears](#)

Conductive hearing loss occurs when there is a problem transferring sound waves anywhere along the pathway through the outer ear, tympanic membrane (eardrum), or middle ear (ossicles). Patients with Kartagener Syndrome can present with chronic infections, specially of the middle ear which can lead to conductive hearing loss.

Situs Inversus (Dextrocardia on Chest X-Ray)

[Inverted-zit and Dexter holding Heart-cards and a Chest-x-ray](#)

Patients with Kartagener Syndrome have Dextrocardia with situs inversus. Situs inversus refers to the mirror-image reversal of the organs in the chest and abdominal cavity. In people affected by dextrocardia, the tip of the heart points towards the right side of the chest instead of the left side. Most patients with this display no sign or symptom and it is not discovered until they have a chest x-ray done.

Screening

Decreased Nasal Nitric Oxide Used as Screening Test

[Down-arrow on Nose of Ox opening Screened-window holding a Nitro-tank](#)

Nitric oxide is continually synthesised in the respiratory epithelium and is upregulated in response to infection or inflammation. Although Kartagener Syndrome patients have chronic infections, nasal nitric oxide in such patients is markedly reduced and is used as a screening test for this condition.