



Arterial Tortuosity Syndrome

a simple twist of fate

Managing treatment of arterial tortuosity syndrome (ATS)

In 2014, Dr Callewaert published important guidelines for treating people with ATS to help provide the best care possible.¹ You can print out this checklist for your next healthcare provider visit.

What you need to know before seeking treatment of ATS

For parents or caregivers

DO try to find a medical center familiar with ATS or similar conditions

DO try to coordinate between different doctors who specialize in different areas of the body for optimal treatment

DO find a doctor willing to direct different doctors involved in managing this condition (pediatrician, generalist, medical, geneticist)

DO find a (children's) cardiologist with experience in this or related conditions

DO consult a dentist to avoid risk of orthodontic complications and make sure the mouth is developing normally

DO find an eye doctor (ophthalmologist) who has experience in connective tissue disorders

DO find an orthopedic surgeon and physiotherapist with experience in connective tissue disorders

DO find a pneumologist (expert in the lungs) with experience in connective tissue disorders

DO encourage children or adults with ATS to participate in swimming and cycling

DO NOT encourage children or adults with ATS to participate in contact sports, competitive sports, repeated isometric exercise (for example weight lifting and scuba diving)

DO NOT use agents that stimulate the cardiovascular system such as decongestants

DO NOT smoke

DO NOT encourage excessive tanning

For women with ATS who are planning a pregnancy or are pregnant

DO seek preconception counseling

DO find a high-care prenatal and postnatal obstetric center with expertise in connective tissue conditions

DO have regular cardiovascular follow-up during pregnancy and within 6 months after birth

DO transition to an antihypertensive such as a beta blocker before conception or as soon as pregnancy is recognized

DO NOT take an angiotensin-converting enzyme inhibitor (ACE-I) or angiotensin II receptor 1 antagonists (ATIIR1) such as losartan, especially in the second and third trimesters

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Reference: 1. Callewaert B, De Paepe A, Coucke P. Arterial Tortuosity Syndrome v2014 Nov 13. In: Pagon RA, Adam MP, Ardinger HH, et al, eds. *GeneReviews*[®]. Seattle, WA: University of Washington, Seattle; 1993-2015.

FOR PHYSICIANS managing treatment of arterial tortuosity syndrome (ATS)

Cardiovascular **! CAUTION: Antihypertensive efficacy has not been established in patients with ATS.**

DO regular cardiovascular follow-up (echocardiography and MRA or CT scan with 3D reconstruction from head to pelvis) starting at birth or at the time of diagnosis

DO consider surgical intervention for aneurysms and focal symptomatic stenosis

BE CAREFUL when reducing hemodynamic stress on arterial walls with ACE-I, or ATIIR1 antagonists. Using these blood pressure-lowering medications in the presence of arterial stenosis (anatomic or functional due to severe tortuosity), and especially renal artery stenosis, may create risk of renal failure

Surgical

DO place stiches without traction and keep in place ~10 days as wound healing may be delayed following surgery

DO use a supporting mesh in surgical repair of hernias to reduce recurrence risk

DO NOT perform surgeries that are medically not strictly indicated (eg, plastic surgery)

Pulmonary

DO lung function tests and imaging if emphysema or asthma is suspected

DO conduct routine follow-up for any pulmonary manifestation such as emphysema and asthma, which are treated symptomatically

DO NOT rely on positive pressure ventilation longer than necessary as it may cause emphysematous changes to progress

Skeletal

DO ask an orthopedist to consider bone densitometry for osteoporosis depending on patient and family history and assess with radiographs if conditions such as scoliosis are suspected

DO evaluate whether surgical stabilization of the spine is necessary

DO evaluate the need for physiotherapy/hydrotherapy in case of joint instability and joint aches

Ocular

DO consult with an ophthalmologist who has experience in connective tissue disorders to evaluate amblyopia, keratoconus, keratoglobus, and corneal thinning

Dental

DO consult on bifid uvula and cleft palate which may cause risk of feeding difficulties

DO consult on potential dental crowding secondary to palatal abnormalities

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