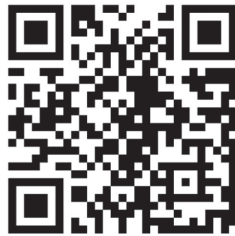




Posterior arch defect

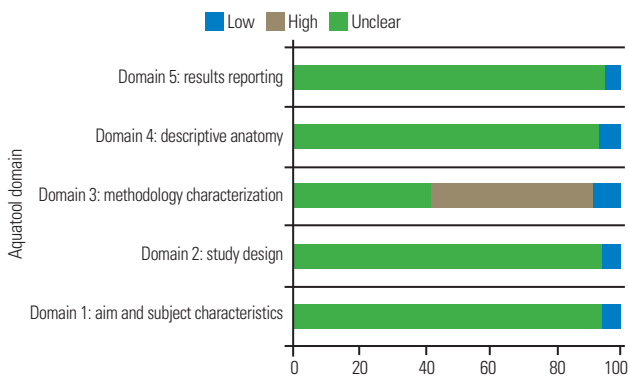


Sided posterior arch defect



Bilateral posterior arch defects associated with anterior arch defect

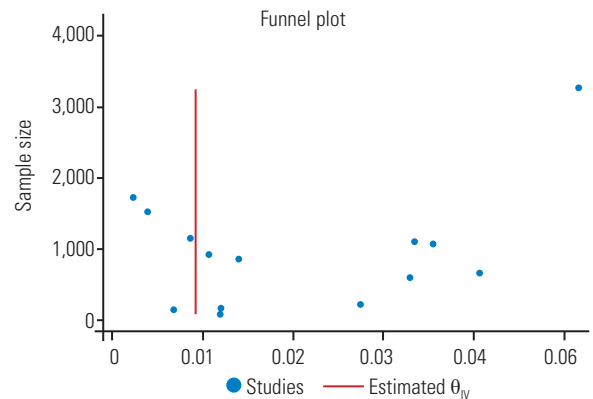
**Supplement 1.** Additional three-dimensional reconstruction models uploaded to Figshare (<https://doi.org/10.6084/m9.figshare.c.6758133>).



**Supplement 2.** Risk assessment results using anatomical quality assessment (AQUA) tool.

**Supplement 3.** All excluded studies and the reasons for exclusion

Study	Reason
Palancar et al. (2020)	Done in Neanderthals
Urrutia et al. (2016)	Defect of lumbar vertebra, not atlas
Koc et al. (2018)	Done in patients with atlas assimilation
Konan et al. (2008)	Done in children with chromosome 22q11.2 microdeletion syndrome
Fuhrop et al. (2015)	Done in in loeys-dietz syndrome patients
Wu et al. (2022)	Done in children
Piatt et al. (2011)	Done in children
Karwacki et al. (2012)	Done in children (ossification pattern)
Giuffra et al. (2016)	Only one case was reported



**Supplement 4.** Funnel plot of sample size versus prevalence.

**Supplement 5.** Results from 48 previous studies according to our proposed classification

Study		Structure(s)	Method	#	Description
Chalmers (1985)	1	Isolated midline AAD	Radiograph	1	25 yr M
Park (2012)	2	Midline AAD with midline PAD	CT	1	22 yr M
Garg (2004)	1	Isolated midline AAD	CT	1	16 yr M
Allam (2015)	2	Midline AAD associated with PAD	CT	1	20 yr M
	3	Left-sided AAD associated with PAD	CT	1	39 yr M
Muthu (2007)	2	Midline AAD with midline PAD	CT	1	21 yr M
Chung (2010)	1	Isolated midline AAD	CT	1	11 yr M
Unlu (2016)	3	Left paracentral AAD with midline PAD	CT	1	7 yr M
Hosalkar (2001)	2	Midline AAD with bilateral PAD	CT	1	5 yr M
Gil (2015)	2	Midline AAD with midline PAD	CT	1	70 yr M
Boron (2021)	2	Midline AAD with midline PAD	CT	1	11 yr F
He (2012)	2	Midline AAD associated with posterior atlanto-occipital fusion	CT	1	46 yr F
Hinai (2021)	2	Midline AAD with midline PAD	CT	2	Unspecified
	2				
Volpe (2021)	2	Midline AAD with midline PAD	CT	2	54 yr M and 25 yr F
	2				
Mishra (2021)	5	Midline AAD with absence of posterior arch (type E)	CT	1	25 yr M
Popat (2008)	1	Isolated midline AAD	CT	1	10 yr M
Rogers (2011)	2	Midline AAD with midline PAD	CT	1	12 yr F
Desgrez (1965)	1	Isolated midline AAD	NA	NA	Unknown As reported in Allam (2015)
Giepel (1955)	1	Isolated midline AAD	NA	NA	Unknown As reported in Allam (2015)
Mace (1986)	1	Isolated midline AAD	CT	1	13 yr M
Glasser (1991)	1	Isolated midline AAD	CT	1	22 yr M
Chambers (1992)	2	Midline AAD with bilateral PAD	CT	1	33 yr F
Van der Velde (1997)	1	Isolated midline AAD	CT	1	19 yr M
Sasaka (2006)	1	Isolated midline AAD	CT	1	75 yr M
Thavarajah (2012)	1	Isolated midline AAD	CT	1	35 yr F
Senoglu (2007)	2	Midline AAD with sided PAD (B)			
Tajima (2010)	1	Isolated midline AAD			
	2	Midline AAD with midline PAD			
Hyun	2	Midline AAD with midline PAD (15)			
	3	Paracentral AAD with left PAD (1)			
	1	Isolated midline AAD (1)			
	4	Bilateral AAD with midline PAD (1)			
Petraglia (2012)	2	Midline AAD with midline PAD	CT	1	19 yr M
Weng (2010)	5	Midline AAD with absence of posterior arch (type E) and odontoid	CT	1	13 yr F
Garg (2004b)	2	Midline AAD with midline PAD and odontoid	CT	1	16 yr M
Osti (2006)	2	Midline AAD with midline PAD and odontoid	CT	1	15 yr M
Hummel (2013)	2	Midline AAD with midline PAD	CT	3	36, 23, and 42 yr M
	2				
	2				
Atasoy (2002)	2	Midline AAD with midline PAD	CT	1	30 yr F
Hu (2009)	2	Midline AAD with midline PAD	CT	1	20 yr M
Osher (2004)	2	Midline AAD with midline PAD	Radiograph	1	23 yr M
Jans (2009)	2	Midline AAD with midline PAD	CT	1	16 yr M
Walker (1995)	2	Midline AAD with midline PAD	CT	1	60 yr M
Cebeci (2016)	2	Midline AAD with midline PAD	CT	1	30 yr F
Salunke (2015)	5	Midline AAD with absence of posterior arch (type E)	MRI	1	42 yr F

(Continued on next page)

## Supplement 5. Continued

Study	Structure(s)	Method	#	Description
Wolf (1997)	2	Midline AAD with midline PAD	CT	1 47 yr F
Marques (2016)	2	Midline AAD with midline PAD and dens hypoplasia	CT	1 62 yr F
Park (2006)	2	Midline AAD with midline PAD	CT	1 56 yr M
Bonnville (2004)	2	Midline AAD with midline PAD	CT	1 22 yr M
Hannah (2009)	2	Midline AAD with midline PAD	CT	1 30 yr F
Friedman (1985)	2	Midline AAD with midline PAD	CT	1 19 yr F
Ulusoy (2017)	1	Isolated midline AAD (with cervical ribs)	CT	1 56 yr F
Martirosyan (2016)	2	Midline AAD with midline PAD	CT	1 16 yr F
Perez-Villina (2002)	1	Isolated midline AAD with fusion with dens	CT	1 13 yr M
Polga (1974)	1	Isolated midline AAD	CT	1 23 yr M
Kuhne (1977)	2	Midline AAD with midline PAD	CT	1 57 yr M
Premph (2002)	2	Midline AAD with midline PAD	CT	29 yr M
Choi (2011)	2	Midline AAD with bilateral PAD	CT	1 46 yr F

O: Normal; A=1: Isolated midline AAD; B=2: AAD with PAD (midline, bilateral, or one-sided); C=3: Sided AAD (midline, one-sided PAD); D=4: Bilateral AAD; E=5: AAD with posterior arch absence. Total no. of studies=52; 1 (n=16), 2 (n=38), 3 (n=3), 4 (n=1), 5 (n=3).

AAD, anterior arch defect; M, male; F, female; PAD, posterior arch defect; CT, computed tomography; NA, not applicable; MRI, magnetic resonance imaging.