Table 4. Common congenital abnormalities associated with DBA syndrome

ORGAN SYSTEM	Frequency, median (range)	FINDINGS
Any type	54.4% (40.6-71.8)	(Including short stature, small for gestational age/intrauterine growth retardation)
Craniofacial and neck	21.6% (14.5-25) *Cleft palate: 4.26% (3.5-5.8)*	Hypertelorism, microcephaly, micrognathia (Pierre-Robin), microtia, broad flat nasal bridge, epicanthus, cleft lip, cleft palate, shorted/webbed neck, Sprengel deformity, Klippel-Feil deformity, low set ears, prominent ears, low hair line, ptosis, mandibulofacial dysostosis (Treacher-Collins syndrome phenocopy)
Cardiac	11.6% (6.9-15)	Ventricular septal defect, atrial septal defect, coarctation of the aorta, tetralogy of Fallot, bicuspid aortic valve, pulmonary stenosis, anomalous venous return, other complex cardiac defects
Thumb and skeletal	18.5% (17.9-19) *Thumbs: 7.6% (6-9.2)*	Thumb (absent, atypical, duplex, bifid, triphalangeal), flat thenar eminence, polydactyly, syndactyly, absence of radial artery, acetabular dysplasia, pectus excavatum
Urogenital	10.7% (6.3-19.5)	Absent or horseshoe kidney, duplicated collecting systems, hypospadias, inguinal hernias
Ophthalmological	Rare	Congenital glaucoma or cataracts, strabismus
Skin	Rare	Café au lait spots, congenital nevi, hemangioma, dermatofibroma
Neurodevelopmental	3% (1.3-4.6)	Learning difficulties, mild to severe developmental delay

Data on frequency are from DBA syndrome registry papers cited in the manuscript.