Case Report

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Radiology Section

First-trimester Detection of Isolated Unilateral Complete Radial Aplasia during Nuchal Translucency Screening: A Case Report

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ABSTRACT

Radial ray defects are congenital upper-limb anomalies involving the radius, radial carpal structures and thumb. We report a 24-year-old primigravida in whom left-sided complete radial aplasia with radial club hand and absent thumb (Bayne-Klug Type IV) was identified at a routine first-trimester nuchal translucency scan at 13 weeks six days of gestation. Detailed sonographic examination showed no other structural anomaly or soft marker. Chorionic Villus Sampling (CVS) was performed at 14 weeks one day for conventional karyotyping, with foetal DNA stored for possible future testing. The couple elected medical termination without awaiting the result. Expulsion followed mifepristone and misoprostol and was complicated by retained placenta requiring emergency suction evacuation and transfusion of one unit of packed red cells. Karyotyping was subsequently reported as normal. Postexpulsion radiography confirmed complete absence of the left radius with a bowed ulna and absent thumb. Chromosomal microarray and exome sequencing were not performed and no autopsy was undertaken; the defect was therefore described as apparently, rather than truly, isolated.

Keywords: Chorionic villus sampling, Congenital abnormalities, Karyotyping, Nuchal translucency measurement, Prenatal diagnosis, Upper extremity deformities

CASE REPORT

A 24-year-old primigravida in a non consanguineous marriage, with no significant medical, surgical, or family history and no reported teratogen exposure, underwent a dating scan at seven weeks one day, confirming a single live intrauterine gestation. A minimally complex right adnexal cyst measuring 2.4×2.3 cm was noted; CA-125 was 18.1 U/mL and the cyst resolved spontaneously by the subsequent examination.

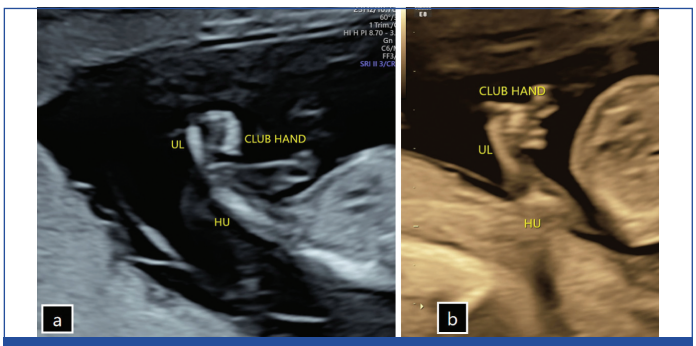
At the routine Nuchal Translucency (NT) scan sonographic parameters and foetal biometry were as summarised in [Table/Fig-1]. During systematic limb survey the left upper limb showed a single forearm bone with fixed radial deviation of the wrist, consistent with complete radial aplasia and radial club hand; the left thumb was absent [Table/Fig-2]. The right upper limb and both lower limbs were normal. Foetal abdominal situs was normal, and both kidneys and the bladder were visualised. No major cardiac abnormality was detected on first-trimester assessment; however, a normal first-trimester cardiac survey does not exclude all cardiac defects, and detailed foetal echocardiography would ordinarily have been recommended had the pregnancy continued. Combined first-trimester screening indicated low risk for trisomy 21. The impression was a left upper-limb radial ray defect (radius absent, club hand), Bayne-Klug Type IV [1,2].

Invasive prenatal diagnosis was recommended. Transabdominal CVS was performed at 14 weeks one day under continuous ultrasound guidance and local anaesthesia using an 18-gauge needle. Chorionic tissue was divided: one aliquot for conventional cytogenetic analysis and a second for DNA extraction and biobanking. Post-procedure foetal heart rate was 154 bpm, with no complications. Baseline maternal investigations were normal except for subclinical hypothyroidism (thyroid-stimulating hormone 5.47 µIU/mL against the laboratory's assay-specific first-trimester reference interval of 0.2-4.3 µIU/mL; free thyroxine1.02 ng/dL, normal), for which levothyroxine 12.5 µg daily was commenced after physician opinion.

Following counselling addressing the sonographic findings, the recognised syndromic associations, the functional implications of

Date / gestational age	Event	Findings
24 Jun 2025 - 7w1d	Dating scan	Single live intrauterine gestation; CRL 1.01 cm; FHR 140 bpm; right adnexal cyst 2.4 × 2.3 cm (CA-125 18.1 U/mL)
11 Aug 2025 - 13w6d	NT scan	CRL 68.5 mm; NT 1.50 mm; nasal bones ossified bilaterally; no tricuspid regurgitation; FHR 152 bpm; cervical length 3.6 cm; placenta right lateral; liquor adequate
	Biometry	BPD 25.8 mm (14w3d); OFD 32.2 mm (14w2d); HC 91.4 mm (14w1d); AC 73 mm (13w6d); FL 16.7 mm (15w0d)

[Table/Fig-1]: Clinical timeline, sonographic parameters and foetal biometry. CRL: Crown-rump length; FHR: Foetal heart rate; NT: Nuchal translucency; BPD: Biparietal diameter; OFD: Occipitofrontal diameter; HC: Head circumference; AC: Abdominal circumference; FL: Femur length.



[Table/Fig-2]: First-trimester ultrasound at 13 weeks 6 days demonstrating left radial aplasia with radial club hand: (a) Greyscale image; and (b) corresponding two-dimensional sepia-filtered image showing a single forearm bone with radial deviation of the hand. HU: Humerus; UL: Ulna.

unilateral radial aplasia and the management options, the couple elected medical termination and chose not to await the cytogenetic result. Mifepristone 200 mg orally was followed 33 hours later by misoprostol 400 µg vaginally, four-hourly. Expulsion occurred, the placenta was retained and emergency suction evacuation was performed, with transfusion of one unit of packed red cells. No immediate postprocedure haemoglobin was recorded. She remained stable and was discharged the following evening. The expelled foetus weighed 35 g. Postexpulsion radiography and gross examination confirmed absence of the left radius with an intact but bowed ulna and an absent left thumb [Table/Fig-3,4]. No autopsy was performed.



[Table/Fig-3]: Postexpulsion (a) Radiography; (b) Gross photograph (anterior view) confirming complete absence of the left radius with a bowed ulna.



[Table/Fig-4]: Magnified postexpulsion radiograph of the left upper limb showing a single bowed forearm bone (ulna), radial deviation of the hand and absence of the thumb ray (arrow).

Cytogenetic analysis of cultured chorionic villi (GTG banding, 450-band resolution; 20 metaphases analysed) was reported on 2 September 2025 as a normal chromosome complement, with no numerical or structural rearrangement. Under the preconception and prenatal diagnostic techniques (Prohibition of Sex Selection) Act, 2003, the sex chromosome constitution was not disclosed. Microarray and exome sequencing were not performed. Post-termination counselling in foetal medicine addressed recurrence risk, genetic testing and future pregnancies.

DISCUSSION

The Bayne and Klug classification grades radial longitudinal deficiency radiographically from Type I (short distal radius) to Type IV (complete aplasia) [1]. The present case was Type IV, with an absent radius, ulnar bowing, radial wrist deviation, and an absent thumb confined to the left-side. Prenatally, the combination of fixed radial deviation of the wrist with a shortened or single forearm bone constitutes the hallmark sonographic sign [2].

Pajkrt E et al., identified 66 foetuses with forearm anomalies; among the 64 with complete follow-up, chromosomal anomalies accounted for 29.7%, genetic syndromes for a further 29.7%, and isolated defects for only 23%. Bilateral lesions were substantially more strongly associated with aneuploidy and syndromes, whereas sonographically isolated unilateral defects showed a low incidence of additional pathology [3]. Paladini D et al., similarly reported that among 100 foetuses with upper extremity malformations, only 12 had an isolated defect, 88 having associated abnormalities in other organ systems [4]. Differential diagnoses include trisomy 18; TAR (thrombocytopenia-absent radius) syndrome; Holt-Oram syndrome (TBX5), associated with cardiac septal defects; Fanconi anaemia, in which a radial ray defect occurs in about half of affected individuals; Roberts syndrome; VACTERL (Vertebral, Anal, Cardiac, Tracheo-oesophageal, Renal, Limb) association; and teratogens such as valproate [5,6]. Here, TAR syndrome was unlikely, as it characteristically involves bilateral radial aplasia with preserved thumbs, whereas this defect was unilateral with an absent thumb. The normal karyotype excluded trisomy 18, and the survey showed no vertebral, renal, gastrointestinal, or cardiac anomaly to suggest VACTERL association. Holt-Oram syndrome and Fanconi anaemia could not be excluded, as neither targeted molecular testing nor chromosome-breakage studies were performed.

Two reports permit direct comparison. Yang PY et al., described six prenatally detected radial ray defects, three unilateral and three bilateral, diagnosed between 12 and 24 weeks; two were trisomy 18, both with additional sonographic abnormalities, one was TAR syndrome and two were isolated, and all six couples elected termination [5]. Kansal M reported a closely comparable case: a Type IV radial ray anomaly with an absent thumb identified at 12 weeks three days in one foetus of a dichorionic diamniotic twin pregnancy in a 24-year-old primigravida; megacystis and a single umbilical artery were noted at the same examination, and interval imaging ten days later demonstrated an abnormal posterior fossa and a suspected hemivertebra, establishing a clinical diagnosis of VACTERL association despite a normal postnatal karyotype [7].

Hillman SC et al., reported a 4.1% higher detection rate with Chromosomal Microarray (CMA) than with conventional karyotyping when the indication for testing was an abnormal ultrasound finding, rising to 10% in their accompanying meta-analysis [8]. In a first-trimester cohort of 338 high-risk CVS pregnancies, Zhang Q et al., detected chromosomal abnormalities by CMA in 16.6% overall and 25.3% of those referred for ultrasound abnormality, with trio whole-exome sequencing identifying clinically significant variants in a further 22.7% (5 of 22) of CMA-negative foetuses with abnormal ultrasound findings [9]. Neither study is radial-ray-specific, but together they show why a normal karyotype cannot be equated with a normal genetic evaluation.

Counselling should address prognosis and postnatal management. In apparently isolated unilateral radial deficiency, survival is unaffected, although wrist alignment, grip and upper-limb function may be impaired.

CONCLUSION(S)

An apparently isolated unilateral complete radial aplasia with radial club hand and absent thumb was detected at the first-trimester NT scan in a foetus with a normal karyotype and no other structural anomaly identified prenatally. Counselling should incorporate both the functional implications of the defect and the limitations of the genetic evaluation actually achieved.

Authors' contribution: VKM: conception and design of the report, sonographic diagnosis and image interpretation, and critical revision of the manuscript for important intellectual content; HAR: acquisition and compilation of clinical data, literature review, and drafting of the manuscript; KK: clinical management of the patient, performance of chorionic villus sampling, sonographic diagnosis

and image interpretation, acquisition of obstetric data, and critical revision of the manuscript; KKR: critical revision of the manuscript for important intellectual content; PN: overall supervision of the work and critical revision of the manuscript for important intellectual content. All authors gave final approval of the version to be published and agree to be accountable for all aspects of the work, ensuring that questions relating to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

Guarantor: HAR is the guarantor of this study. She accepts full responsibility for the work and the conduct of the study, had access to the data, and controlled the decision to publish. Informed consent was obtained from the subject involved. Appropriate consent has been obtained for all images presented.

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