# Neonatal imaging: Diagnosis

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We congratulate Dr Saifullah Khalid, Department of Radiodiagnosis, Jawaharlal Nehru Medical College, Aligarh Muslim University, Aligarh, Uttar Pradesh, India, for her excellent diagnosis, for which she receives the award of R1 000 from the RSSA. Notable second-place entries were made by Drs Aadil Ahmed, Samuel Manikkam, Jacqueline du Toit, Phil Harries-Jones and Shaun Scheepers. Drs Misser *et al.* elaborate below on the images and findings. Please refer to page 116 of the August 2013 issue of the *SAJR* (http://dx.doi.org/10.7196/SAJR.934) for the presenting details and the investigative images.

## Diagnosis

### Imaging and findings

This first set of radiographs was taken of a neonate born at 33 weeks' gestation. Birth weight was 1.5 kg and the initial Apgar scores were satisfactory. Respiratory distress and bile-stained nasogastric returns were noted post-delivery. Fig. 1 is a plain abdominal radiograph showing the umbilical venous line, right femoral peripheral line to the IVC and a nasogastric tube with tip in the distended pyloric antrum. 'Double bubble' configuration of the stomach and first segment of the duodenum is noted. The rest of the abdomen is gasless. Incidental note is made of the hemivertebra of the tenth dorsal level with missing ipsilateral left tenth rib. Slight angulation of the sacral tip to the right is also identified. The lateral shoot-through (Fig. 2) confirms the bowel abnormality and the apparent anomaly of the forearm bones with club hand configuration. This is confirmed on the babygram image (Fig. 3) where the left radius and thumb is noted to be absent (type IV radial club hand deformity). In addition, the moderate cardiomegaly and plethoric appearance of the lung fields raise suspicion for an intracardiac left-to-right shunt (e.g. atrioventricular septal defect/AVSD) or patent ductus arteriosus (PDA). The gastroduodenal distension, D10 hemivertebra, gentle rightward scoliosis, absent left tenth rib and sacral angulation are all again noted. Fig. 4 reveals a harlequin deformity of the orbits, which is associated with craniosynostosis of the coronal suture or plagiocephaly.

Clinical examination of the child noted the abnormality of the left forearm and wrist with club hand deformity, a sacral pit and a systolic cardiac murmur. There was no marked brachicephaly or other cranial deformity. The respiratory distress was managed as for surfactant deficiency disease, a surgical opinion was sought regarding the bowel abnormality, and a full cardiac assessment was arranged. The echocardiogram revealed three left-to-right shunts including an ASD, ventricular sepal defects (VSD) and a large PDA. The duodenal atresia was successfully corrected and the post-operative period was uneventful. Further investigation included chromosomal studies, which were negative for major chromosomal abnormalities. The combination of all the clinico-radiographic findings made VACTERL association (VA) (Vertebral anomalies, Anal atresia, Cardiac defects, Tracheoesophageal fistula and/or Esophageal atresia, Renal anomalies and Limb defects) the most likely diagnosis. Fanconi anaemia is an important diagnosis to exclude by relevant testing. The apparent harlequin deformity was probably incidental, but may be a feature of Baller-Gerold syndrome, which has a few overlapping features with VA. In the last month, the baby had successful cardiothoracic surgery for correction of the septal defects and is currently recovering well. Further management includes future orthopaedic mangement of the left upper limb deformity.

## Discussion

VA is the expanded acronym for the nonrandom association of multiple birth defects originally described as the VATER syndrome.[1] The term 'association' is thought to be more appropriate than a syndrome as the features are not pathogenetically linked but rather tend to occur more frequently in such a group than in the general population. The abnormalities are predominantly of structures derived from the embryonic mesoderm. Table 1 outlines the various systemic abnormalities that are associated with the VACTERL constellation. A diagnosis of VA is made if at least three of the clinical associations are present.

### Genetics

Most cases of VA are sporadic. The condition rarely occurs twice in a family. The incidence is estimated at 1.6 in 10 000 live births. No specific gene locus has been identified, but there are several genes implicated in this condition. Sonic hedgehog gene (SHH), for instance, is regarded as a keystone sequence, especially in those patients with oesophageal atresia and anorectal malformations.[2] SHH encodes for an intracellular signaling protein vital for embryogenesis. Early disruption of the mesodermal differentiation in utero at the first 5 weeks has been suggested as the basis for the nonrandom association of VA.[3]

In addition, VA is known to occur more frequently in infants born to diabetic mothers.[4] Microdeletions of the FOX gene cluster at chromosome 16, long arm deletions of chromosome 13 and 12 as well as trisomy of chomosome 18 have been implicated among other gene defects.

**Table 1. Spectrum of systemic associations related to VACTERL association**

***V***ertebral
- Hemivertebrae
- Fused vertebrae
- Hypersegmentation
- Sacral anomalies
- Scoliosis
- Sternal anomalies

***A***norectal
- Persistent urachus
- Anal atresia + fistula

***C***ardiovascular
- Ventricular septal defects
- Patent ductus arteriosus
- Tetralogy of Fallot
- Transposition of the great arteries
- Right/double aortic arch
- Coarctation of aorta
- Dextrocardia
- Single umbilical artery
- Anomalous pulmonary venous drainage
- Left superior vena cava

***T***racheo-oesophageal fistula

***E***sophageal atresia

***R***enal anomalies
- Agenesis/dysgenesis
- Ectopia
- Horseshoe kidney
- Ureteral/urethral
- Hydronephrosis
- Vesico-ureteric reflux
- Posterior urethral valves
- Pelvi-ureteric junction anomaly

***L***imb defects
- Radial dysplasia/aplasia
- Hypoplastic/triphalangeal thumb
- Polydactyly
- Syndactyly
- Radio-ulnar synostosis

Other associated defects
- Gastro-intestinal abnormalities
- Ear anomalies
- Potter facies
- Wide suture/large fontanelle
- Cleft palate
- Short stature
- Failure to thrive
- Pancreatic heterotopia
- Spinal dysraphism/tethered cord
- Genital anomalies

## Vertebral defects

These are probably the most common association in VA. The spectrum includes segmentation anomalies such as hemivertebrae, fused vertebrae or hypersegmentation. Any combination of vertebral and other skeletal anomalies is seen, especially rib and sternal abnormality. Cord tethering and sacral abnormalities also occur more frequently. The vertebral defects rarely account for mortality in these patients. Recently, sonar has been introduced as a useful modality in the assessment of vertebral defects.

## Anorectal malformations

The presence of an anorectal abnormality is clinically apparent in the first few days of life and usually requires early surgical correction. Some children will require repeat corrective or reconstructive surgical procedures.

## Cardiac defects and vascular anomalies

In many series of VA, congenital cardiac abnormalities prove to be the most common associated defect. Intracardiac shunts, particularly VSD, are seen frequently. In most patients, the VSD is generally one of several cardiac defects present, rarely found in isolation. The single umbilical artery was noted to be present more commonly in VA, and it was suggested by Temtamy *et al.*[4] shortly after the initial description that V in VATER should include vascular, in addition to vertebral abnormalities.

## Tracheo-oesophageal fistula and atresia

These foregut abnormalities occur variably in 50 - 80% of patients with VA.[5] Antenatal sonar diagnosis of polyhydramnios or absence of the stomach fluid echo should alert the obstetrician to look for other possible congenital abnormalities. Early post-partum surgery is indicated after the subtype of foregut anomaly is determined. Early and late neonatal complications including chest infections are an important cause of co-morbidity.

## Renal abnormalities

These may be less apparent than other components of VA, and dedicated imaging of the renal tract may be required to elucidate such anomalies. Structural renal malformations may be a significant cause of morbidity, particularly recurrent infections and premature renal failure.

## Limb defects

Several limb malformations have been added to the list of possible appendage defects. The classic description included radial ray anomalies also known as radial club hand. Classifications of this deformity originally described with gradation of mild to severe radial aplasia have been revised with several additions, including recent inclusion of a fifth sub-type of absent radius with associated deficiency of the humerus and variable involvement of carpal bones.

## Differential diagnosis

There is considerable overlap between the various syndromes; subtle findings on clinical examination, imaging and laboratory testing will enable a final diagnosis. Proper diagnosis aids further adequate genetic counseling of families. Table 2 delineates the main distinguishing features in the major differential diagnoses, which include Fanconi anaemia, Feingold syndrome, Charge syndrome, DiGeorge syndrome (or 22q11.2 deletion syndrome), oculo-auriculo-vertebral syndrome, Currarino syndrome and VACTERL H. In South Africa, the relative increased incidence of Fanconi anaemia in our population of Afrikaner descent must alert the clinician to the relevant testing in any patient with features of VA owing to the considerable overlap of features, especially the radial ray abnormality. The recessive inheritance pattern, propensity for malignancies in early adulthood and shortened lifespan of patients with Fanconi anaemia warrants early diagnosis of this entity, as genetic counselling is increasingly being offered to affected families.

**Table 2. Key features distinguishing the main differential diagnoses of VACTERL association**

| Syndrome | Features distinct from VA |
|---|---|
| Fanconi anaemia | Haematologic abnormalities, especially megaloblastic anaemia<br>Pigmentation abnormalities, especially hyperpigmentation or Café au lait spots<br>Propensity to develop malignancies |
| Alagile syndrome | Biliary duct abnormality<br>Eye anomalies<br>Facial abnormality |
| Charge syndrome | Choanal atresia<br>Colobomata<br>Growth impairment<br>Ear anomalies |
| Feingold syndrome | Digital phalangeal abnormalities<br>Microcepahly<br>Cognitive impairment<br>Typical facial appearance |
| Oculo-auriculo-vertebral syndrome | Microtia<br>Hemifacial microsomia<br>Neurocognitive impairment |
| Deletion 22q11.2 (DiGeorge) syndrome | Hypocalcaemia<br>Learning difficulties<br>Immune dysfunction |
| VACTERL–H | Hydrocephalus |
| Baller-Gerold syndrome | Craniosynostosis<br>Polymicrogyria, mental retardation |

## Mangement and prognosis

The management of patients with VA depends on the attendant components of the spectrum present at birth. Major life-threatening malformations require urgent neonatal surgical attention. Orthopaedic treatment of limb abnormalities and corrective surgeries for cardiac malformations and scoliosis may be planned/staged in several surgical procedures. The vast development in specialised neonatal and post-surgical ICU facilities has resulted in a better overall prognosis in the managed VA child. The subsequent long-term prognosis is variable and highly dependent on the severity of the individual components in the patient. In addition, each patient will have to endure a further list of complications, related to the component encountered, throughout life. Particular note must be made of the normal neurocognitive functioning in patients with VA. This is a significant positive feature. Some patients may have a normal lifespan without life-threatening issues, but the vast majority will require close surveillance for complications owing to significant associated morbidity.


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# Body imaging

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A 38-year-old woman presented with a painful left paramedian infra-umbilical abdominal wall mass. She was a keen athlete and otherwise well. The following ultrasound images and MRI sequences (after gadolinium administration) were obtained. Describe the relevant imaging findings and formulate the most appropriate clinical diagnosis.

Please submit your response to misser@lakesmit.co.za not later than 15 January 2014. The winning respondent will receive a R1 000 award from the RSSA. A detailed diagnosis and discussion will be presented in the next issue of the *SAJR*.

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# QUIZ CASE

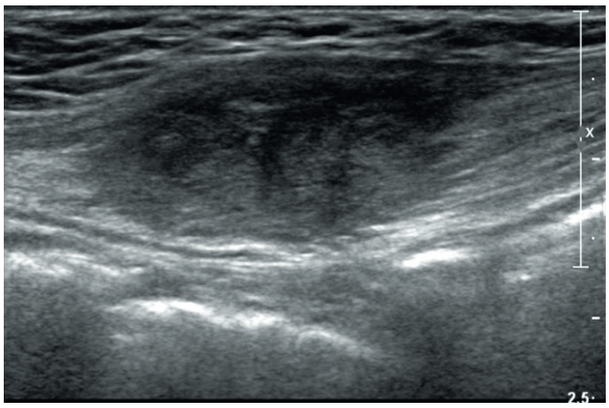

*Fig. 1. Sagittal B-mode sonar image.*

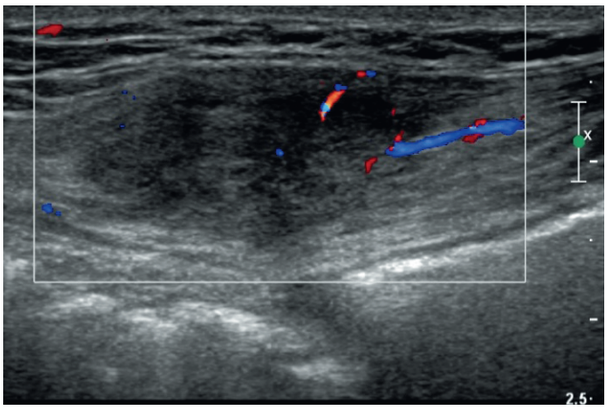

*Fig. 2. Sagittal colour duplex sonar image.*

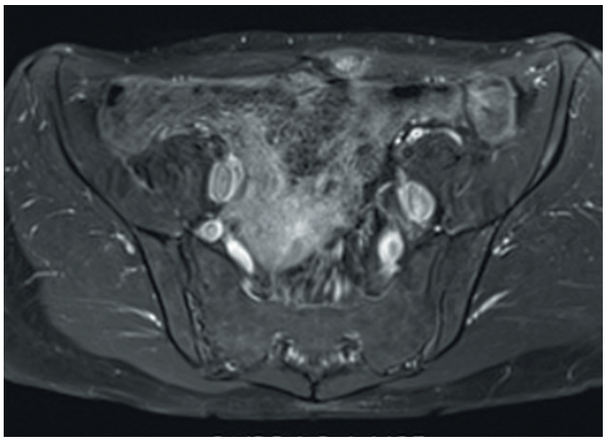

*Fig. 3. Axial post-Gd T1-weighted image through lesion.*

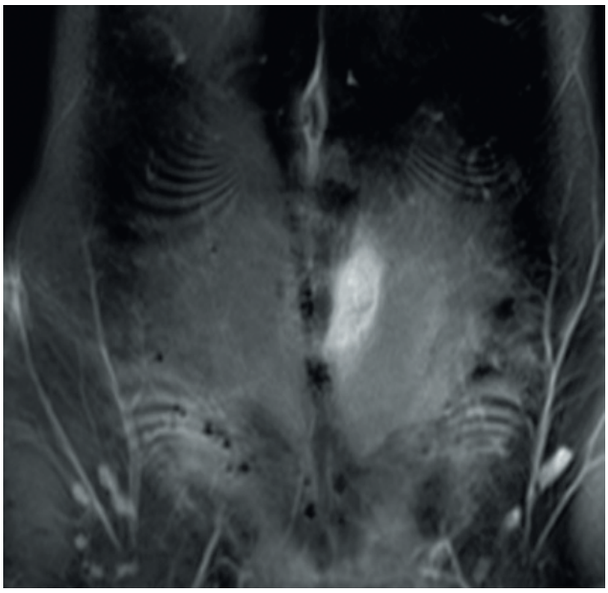

*Fig. 4. Coronal post-Gd T1-weighted image.*

# RADIOACTIVE NEWS

## International Day of Radiology – 8 November 2013

A vital part of medical care since X-rays were discovered in 1895 by Wilhelm Röntgen, radiology and the new technologies developed around it have hugely facilitated better care as well as minimally invasive surgery. In recognition of radiology's impact, radiological societies across the world celebrated the second International Day of Radiology (IDoR) on 8 November 2013.

The IDoR was established to build greater awareness of the value that radiology contributes to patient care, and to improve the understanding of the vital role that radiologists play in the healthcare continuum. Medical imaging is one of the most exciting and progressive disciplines in healthcare and a field of great activity in technological and biological research.

The day is a joint initiative of the ESR, the RSNA and the ACR, with the full co-operation and involvement of the International Society of Radiology (ISR), as well as umbrella organisations on all continents, including the Asian Oceanian Society of Radiology (AOSR), the Colegio Interamericano de Radiología (CIR), the Royal Australian and New Zealand College of Radiologists (RANZCR), and the Radiological Society of South Africa (RSSA – which also represents neighbouring countries). The European Federation of Radiographer Societies (EFRS) also supports IDoR 2013.

Information about the IDoR, including booklets about oncological imaging and the history of radiology, is on the day's official website at http://www.internationaldayofradiology.com. One can also keep up to date with the latest news by liking the IDoR Facebook page.

## AOSPR Congress report-back

*Nishentha Govender*

Firstly, I wish to thank the RSSA CME Association for the generous sponsorship that afforded me the opportunity to attend the 13th Asian & Oceanic Society for Paediatric Radiology (AOSPR) Congress in Hong Kong from 14 - 15 September. It was held during the auspicious moon festival period, which is a wonderful cultural celebration, and was addressed by more than 30 renowned speakers, experts in their respective fields. The congress was not only an opportunity for new ideas and scientific knowledge to be shared, but also fostered collaboration among different specialists and healthcare professionals in pursuit of better healthcare for children worldwide. The organising committee put together a most comprehensive programme which dealt with a broad scope of paediatric radiology. The topics ranged from paediatric emergencies to subspecialist medical advances. I am always astounded at the advances in radiology and how the boundaries of science are being challenged. Yet another example of this was covered in the interesting presentation by Giles Boland, Professor of Radiology at Harvard Medical School, on 'Precision imaging in the era of personalised medicine', in which he discussed the use of imaging biomarkers in oncology and clinical trials.

The AOSPR provides radiologists with a wonderful opportunity to make new friendships and share experiences with each other. I had the privilege of delivering both an oral and a poster presentation concerning our South African-based research papers.

The AOSPR strongly encourages submission of abstracts and participation in the congress. I would advise registrars and interested researchers to take advantage of this opportunity to share their scientific contributions to an international audience. The next AOSPR meeting will be held in Melbourne and is predicted to be even better.

Thanks again to the RSSA for this much-appreciated, great experience.

## Report on the 37th ESNR Annual Meeting

*Sally Candy*
*Head of Neuroradiology and MRI*
*Groote Schuur Hospital and UCT*

The 37th European Society of Neuroradiology/21st advanced course in Diagnostic Radiology and 5th course in Interventional Neuroradiology took place between 29 September and 1 October at the Conference Centre, Westend Campus of the Goethe University, Frankfurt. This well designed and modern venue served the conference well, with its emphasis on state-of-the-art neuro-intervention and diagnostics.

Professor Anton Valavanis gave a sparkling review of the microcirculation and collateral vascular supply of the brain that provided the framework for the subsequent talks on chronic ischaemia, cerebral haemodynamics nd the role of the many advanced imaging techniques now available to the neuroradiologist. Susceptibility weighted imaging, MR perfusion and arterial spin labelling featured prominently. Reference was made to the disappointing results of intra-arterial thrombolysis, but the beneficial role of reperfusion in clinical post-stroke outcome was emphasised. Prospective studies using technologically advanced stent retrievers in 'bridging' treatment are ongoing, and the proponents of this form of acute stroke management are optimistic.

Despite the improvements in diffusion weighted imaging (DWI) and diffusion tensor imaging (DTI) in tumour and infection of both the brain and the spine, accurate prediction of tumour grade and histology are still not within reach. A group in Spain presented preliminary findings of DTI in quantitative assessment of the degree of degeneration in intervertebral discs.

Dynamic fetal imaging now allows us to assess foetal cardiac and gastro-intestinal abnormalities to maximise pregnancy outcome and to plan delivery management and 'exit' strategy.

7 Tesla imaging is now a clinical reality. Professor Pham (Germany) elegantly demonstrated how high-Tesla magnets offer exquisite visualisation of the internal architecture of previously impenetrable peripheral nerves and plexuses. Professor Rovira (Spain) outlined the advantages of high-strength 3D FLAIR, white matter-attenuated TFE, T2* GRE and SWI imaging in improving the conspicuity of demyelinating plaques in the cortex of patients with multiple sclerosis.

Functional imaging is finding use in the imaging of pre-clinical dementia and pain perception and control. The prize for futuristic possibility, though, must go to Dr Gobel of Maastricht University, the Netherlands, who expanded on the use of complex functional MR feedback loops to enable 'locked-in' patients to communicate.

Anne Osborn gave two outstanding review lectures on Tumours in Epilepsy and The Spectrum of Lymphomatoid Disorders. Some of us in South Africa will have been fortunate enough to have had a sneak preview of this lecture on a webinar arranged by the RSSA earlier this year.

Congratulations to the President of the Congress, Prof. Dr Friedhelm Zanella and to Prof. Turgat Tali, ESNR President, for a well-organised and stimulating meeting.

## WFNRS planning meeting for the XXth Symposium Neuroradiologicum

*Sally Candy*
*Head of Neuroradiology and MRI*
*Groote Schuur Hospital and UCT*

The meeting, chaired by Professor E Turgat Tali, was the second meeting of the organising committee for the Symposium Neuroradiologicum to be held in Istanbul, Turkey, from 8 - 12 September 2014. The first planning meeting was held in San Diego at the ASNR earlier this year. Representatives from all over Europe attended. Drs Candy, Kilborn and Janse van Rensburg represented the subgroups of SASNI and SASPI RSSA at the meeting, and indicated their willingness to review scientific abstracts and to chair sessions at the prestigious symposium, where several parallel sessions will cover the topics of stroke, head and neck, paediatric diagnostics, diagnostic and interventional spine, intravascular intervention and adult imaging.

A decision was made at the meeting that the poster session will be conducted entirely electronically, and that the parallel scientific oral presentations will be preceded by a named speaker to provide coherence and to improve attendance.

The website for abstract submission opened on 11 September this year and will close on 31 March 2014. Delegates will be notified of abstract acceptance on 30 April 2014. On-line registration opens on 22 November 2013. The website URL is http://www.wfnrs2014.com/en/