

# Polythelia and supernumerary cervical and thoracic vertebrae

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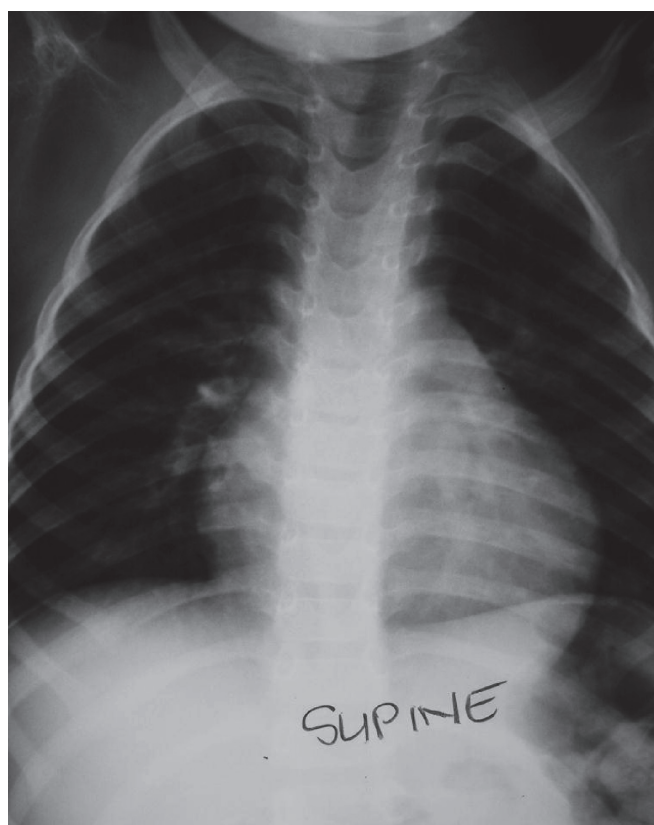
We present the case of an 11-year-old boy with an elongated neck and chest. Both the cervical and the thoracic spine carried one supernumerary vertebra. This was accompanied by symmetrical polythelia (i.e. additional nipples). To our knowledge this is first publication of a case presenting with this collection of features.

## Case report

An 11-year-old boy presented to the Trauma Unit at Red Cross Hospital after sustaining an injury to the neck area. Radiographs of the cervical spine showed no signs of trauma. However, it was noted that the cervical spine comprised 8 vertebrae instead of the usual 7 (Fig. 1). In addition, the thoracic radiograph demonstrated 13 vertebrae (Fig. 2). On physical examination the patient was also found to have two sets of nipples, approximately 4 cm from each other (Fig. 3).



**Fig. 1.** Lateral cervical radiograph, demonstrating the 8 vertebral bodies.



**Fig. 2.** Anteroposterior thoracic radiograph, demonstrating the 13 vertebral bodies, all with ribs attached.



**Fig. 3.** Ventral aspect of the proximal anterior chest cage, demonstrating the two sets of nipples.

## Discussion

Extra vertebrae in the neck region are rare, and the combination of symmetrical hypertelism and elongated chest has not, to our knowledge, been documented before.

## CASE REPORT

The vertebral bodies are formed from the 4th week onwards as a result of migration of cells from the sclerotome regions of the somites in the ventromedial, ventrolateral and dorsal direction. Chondrification takes place from the 6th week onwards.<sup>1</sup>

Mammary buds begin to develop during the 6th week as solid downgrowths of the epidermis into the underlying mesenchyme.<sup>2</sup> Supernumerary nipples or polythelia are developmental abnormalities located along the embryonic mammary lines. This is the most common form of accessory breast tissue, found in 0.22 - 5.6% of people, depending on various factors such as gender, ethnic group and geographical area.<sup>3</sup> They are usually sporadic and rarely familial and may be associated with kidney anomalies.<sup>4</sup> They may be found anywhere along the milk line but most often appear below the usual site of breast placement. They are occasionally symmetrical or multiple, but are most often solitary.<sup>5</sup>

Polythelia and segmental costovertebral malformations have been described in single case in association with neural tube defects.<sup>6</sup> However, a case such as ours, presenting with supernumerary cervical and thoracic vertebrae in addition to polythelia, has not to our knowledge been reported previously.

The case could be described as a long neck and an elongated trunk. The combination of the extra thoracic vertebral body and the doubling of the nipples suggest that the duplication occurred at the level of the 4th thoracic vertebral body, occurring somewhere between the 4th and 6th week of intrauterine development.

### References

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### How Bio-Rad contribute to society

#### Newborn screening

Bio-Rad Laboratories has been involved in the screening of haemoglobin disorders since the introduction of the Variant™ "Classic" over ten years ago. Since that time, Bio-Rad Laboratories has established a very strong reputation as the supplier of choice for the screening of such haemoglobin disorders as β-Thalassaemia, alpha Thalassaemia and Sickle cell disease.

These disorders are genetically inherited and may present as either largely harmless carrier states or full blown diseases. Carriers can pass on the genetic information to their offspring, giving rise to either further carriers or subjects that suffer the effects of the disease proper.

These diseases have generally been considered to be associated with peoples from the Caribbean, Africa, Gulf States, Pakistan and Far Eastern countries. However, this is no longer the case due to global migrations and inter-cultural marriages.

In respect of Sickle cell disease, so named due to the sickle shape of the red blood cells, the carrier state is believed to prevent infection by the malarial parasite thereby conferring partial immunity to the carrier. However, the full blown disease state can be a fatal condition if left untreated.



Red blood cells, showing one with characteristic sickle shape

By using a small amount of blood taken from a heel prick onto a special filter paper (Guthrie card), it is readily possible to screen babies for a number of treatable but life threatening diseases. Very many countries already offer a "Newborn Screening" program for diseases such as congenital hypothyroidism (*inadequate thyroid hormone production*) and Phenylketonuria (*missing an important amino enzyme, can cause severe mental retardation*) but not Sickle cell disease.



Bio-Rad Vnbs newborn screening analyser

Starting first by working with the state of California, Bio-Rad Laboratories has developed a specific HPLC based analyser, the "Newborn Haemoglobin Screening System" (NHS), which enables laboratories to rapidly screen every baby born for Sickle cell disease and thereby allow for prompt baby treatment and parent counselling.

The UK government decided upon a National Newborn Screening Program for Sickle cell disease in 2001. This represented a brand new governmental initiative, which has led to every baby born in England (around 500,000) being screened for this disease. After three years of liaison between Bio-Rad Laboratories and the UK screening committee, we have successfully produced a modification of the "NHS", called the Variant™ newborn screening system (Vnbs). I am very proud to report that this device is being used for over 90% of all the screening in England and generates revenues of over €1,000K per year. Following on from the success in England, and based on a desire to

offer improved population healthcare, we now see further opportunities as other countries establish newborn Sickle cell disease screening initiatives.

The Dutch government has already agreed that it should be offering this type of screening but have yet to "go live". We have an ongoing Vnbs evaluation being conducted between Leiden Univer-



Obtaining a sample from a newborn is simple

sity Hospital and the RIVM screening laboratory, which is looking very promising.

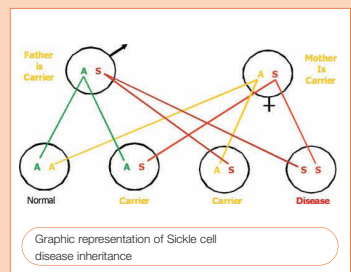
Similarly, the French government wishes to start a screening program for this disease and we are working very closely with the Henri Mondor Hospital who are also evaluating a Vnbs. I wish Sophie Bui and her colleagues every success in this project.

Meanwhile in Brussels, Dr. Gulbis is working to demonstrate the advantages of newborn screening for Sickle cell disease and we at Bio-Rad are working just as hard to convince all concerned that the Vnbs is the instrument of choice.

As a Northern European product manager, I am required to visit our colleagues in South Africa, Kenya and Tanzania. Over the last three years we have been working hard at getting haemoglobin screening established in these countries. Slowly our work bears fruit and we now have a steadily growing base of screening laboratories in South

Africa. Two years ago, it was my pleasure to help our South African team install a Variant™ and train staff at the Malaria Research Unit, in Kenya, where the assessment of Sickle cell disease is now a critical part of their malaria studies.

Most recently we established a relationship with both the Wellcome Research laboratory in Kenya and Muhimbili University Hospital in



Graphic representation of Sickle cell disease inheritance

Tanzania where pilot studies into the incidence of Sickle cell disease and population mortalities have been launched. The doctors and lab workers are using two Variants to help them understand the relationship between Malaria and Sickle cell disease.

As we enter into our new phase of development as a European region, we can clearly say that Bio-Rad Laboratories is truly a culturally diverse group providing social improvements for not only established first world countries but also to those areas that continue to meet their healthcare challenges. I think it is safe to say we will all develop and grow together.

Jon Strotton  
Northern European Product Manager  
Diabetes and Haemoglobinopathies