Eur J Anat, 7 Suppl. 1: 1-3 (2003)

Meaning and clinical interest of the anatomical variations in the 21st century

J.R. Sañudo[1], R. Vázquez[2] and J. Puerta[3]

1- Unitat d'Anatomia i Embriologia, Facultat de Medicina, Universitat Autónoma de Barcelona, Spain

2- Departamento de Anatomía e Histología Humanas, Facultad de Medicina, Universidad de Salamanca

3- Departamento de Anatomía y Embriología Humana I, Facultad de Medicina, Universidad Complutense, Madrid, Spain

The history of the concept and contents of the human anatomical variations is the history of the anatomy itself, or, more accurately, the history of the search for the establishment of the *canon* of the normal structure and composition of the human body. Normal means "*within the normal range of variation*" (Moore, 1989).

Singer credits Eustachius (1520-1574) with the introduction of the study of human anatomical variability. However, there is no doubt that his works actually postdate those of Vesalius, *Humani Corporis Fabrica* (1542), which are full of references to anatomical variations of bones, muscles and vessels; and of course those of Galen and another ancient anatomists (Vesalius, 1542; Straus and Temkin, 1943; Singer, 1956; Hast and Garrison, 2000).

To recognise anatomical variations it is necessary to previously establish the normal pattern of the human body and name its structures. Galen initiated this effort centuries before Vesalius, based on his clinical practice and dissection of apes, dogs and other mammals (Singer, 1957). Galen's anatomy survived until Vesalius' time as a dogma for medical education and practice (Straus and Temkin, 1943; Singer, 1956). He named most of the structures of the human body and encouraged to do dissection to avoid misinterpretation and to increase knowledge (Straus and Temkin, 1943; Singer, 1956). Vesalius, following Galen's advice, used the dissection as the main tool to understand the structure of human beings, thus initiating the era of modern Anatomy.

Vesalius, like Galen, noticed several individual variations, hence the abundance in his work of expressions which can be translated into "always", "usually", "frequently", "more frequently", "most frequently", "sometimes", "not always", "rarely", "relatively rarely", "much more rarely", and "very rarely" (Straus and Temkin, 1943). The adaptation of these terms to our actual background could lead us to consider that Vesalius established variations based on a statistical criteria. However, Vesalius made some erroneous interpretations; for instance, he reported a sacrum bone consisting of five vertebrae as a variation, being a six-vertebrae bone the norm; or he considered as natural a skull with a high forehead, being the other type unnatural (Straus and Temkin, 1943; Hast and Garrison, 2000). Some of the Vesalius' misinterpretations were due to his not complete riddance of Galen's Philosophical and theological influence (Straus and Temkin, 1943).

Several centuries and dissections were necessary to establish the concept of normality, abnormality and variation of the human body. This knowledge based upon the works of many biologists, clinicians and anatomists flourished towards the end of the 19th century and beginning of the 20th, and continues until now with the publication of books and articles focused on anatomical variations (Bergman et al., 1988; Bergman et al., 2002).

The terms "anomaly" or "abnormal" were not used by Vesalius. However, it is commonly known that individuals of the same species are never exactly alike. Both the body as a whole

Correspondence to:
Dr. J.R. Sañudo. Departamento de Anatomía y Embriología Humana I, Facultad de Medicina, Universidad Complutense, 28040 Madrid, Spain.

and its internal organs and parts show certain flexibility of size, form, structure and position. Such fluctuation within a commonly experienced range is considered as "normal variations", nevertheless, any departure beyond these limits is classified, specially of the more extreme types, as *anomalies* or *malformations* (Arey, 1940). Anomalies or malformations are considered as synonymous for structural abnormality (Holmes, 1976). The structural abnormalities are caused by aberrant processes of development during the formation of a particular structure that could be caused by genetic, chromosomical or environmental influences or by a combination of them (Arey, 1940).

Anomalies have been arbitrarily classified in minor or major anomalies (Marden et al., 1964; Stevenson and Hall, 1993), being the differences between those types functional and cosmetic. While the consequences of major anomalies are a common cause of illness, handicapping conditions and death, minor anomalies do not have medical consequences for the patient (Marden et al., 1964; Holmes, 1976; Stevenson and Hall, 1993). Therefore, minor anomalies sometimes lack clear distinction with variations.

The incidence of minor anomalies has been reported to range between 7% to 41% while that of major anomalies lies between 2% and 3% (Stevenson and Hall, 1993).

The term of minor anomalies has been used for describing morphological defects that could be observed externally: flat occiput, Darwinian point, hypoteloism, cleft uvula, microglossia, sacrum dimples, pigmented spots, showl scrotum, cubitus valgus, prominent heel, etc. (Marden, 1964; Stevenson and Hall, 1993).

The apparent lesser value of minor anomalies may be overlooked by the physician, even though it may well represent a valuable clue to altered embryonic development. Infants free of minor defects have low incidence of major anomalies (1%); infants with one minor defect have a 3% risk of major defects; those with two minor defects have a 10% risk of major anomalies, and those with three or more minor defects have a 20% risk of a major defect (Méhes, 1983; Marden et al., 1964). Therefore, the finding of several minor anomalies in a baby might alert the physician to the existence of other defects of a more serious nature (Marden et al., 1964).

A minor anomaly is distinguished in statistical terms from a variation because it could be associated with a major one and occurs in 4% or less of the infants of the same racial group (Holmes, 1976). This is a fourfold greater incidence than the 1% required for a human polymorphism (Stevenson and Hall, 1993).

Thus, based on the aforementioned reasons, we could consider that the term malformation or anomaly is applicable when the structural change has a negative influence under normal circumstances on the function of the organ. In contrast, variations generally have no effect on the function of the organ under normal circumstances, though even a harmless variation can have negative effects under certain circumstances (Lippert and Pabst, 1985).

Once the meaning of the term variation has been established, it is necessary to comment that terms like anomaly, abnormal or aberrant to refer a morphological variation must be avoided. Today it is not surprising to find papers reporting variations that used the mentioned terms.

One question that could be asked today is if publication and study of anatomical variations has sense in the medicine and biology of the 21st century, or is, on the contrary, an exhausted topic (William and Humpherson, 1999). The answer is relatively easy: medical progress needs a more accurate knowledge of the variability of the human morphology to improve diagnosis and therapeutic performance. In other words the advance in the new imaging techniques (echography, MRI, CT, endoscopy, etc.), surgery (reconstructive, minimal invasive surgery, etc.) and other areas, has opened a new field of research for the descriptive anatomy that we consider promising (Jones et al., 2002). On the other hand, anatomical variations represent an embryological and comparative background for medicine and biology in order to understand the morphological aspect of the human body and its related structures. Finally, there is a point in commenting that a recent paper states that about 10% of clinical malpractice is due to the ignorance of the anatomical variations (Cahill and Leonard, 1999). It is not surprising that anatomical variations not only have not disappeared from the medical and biological background but also have also been enclosed among the main aims to be considered in medical curricula in Netherlands and USA (Educational Affairs Committee of the AACA, 1996; Griffioen et al., 1999).

Anatomical variations will always have a place in the medical or biological background; however, we must care the quality of the papers by means of careful review of previous publications and intent of explanation of its possible origin, without forgetting its clinical interest, prior to publishing a contribution (Fontaine, 2001a, b).

As conclusion, we would like to say that the publication of this special issue of the European Journal of Anatomy must thank the contributors of the volume and encourage anatomists to publish articles related to human variations in the century of the molecular biology and genetics.

References

Arey LB (1940). *Developmental anatomy*. W.B. Saunders Company, Philadelphia and London.

BERGMAN RA, THOMPSON SA, AFIFI AK and SAADEH FA (1988). *Compendium of human anatomic variations*. Urban & Schwarzenberg, Baltimore-Munich.

BERGMAN RA, AFIFI AK and MIYAUCHI R (2002). Illustrated encyclopaedia of human anatomic variations. http://www.uh.org/Providers/Texbooks/AnatomicVariants/AnatomyHP.html.

CAHILL DR and LEONARD RJ (1999). Missteps and masquerade in medical academe: clinical anatomists call for action. *Clin Anat*, 12: 220-222.

EDUCATIONAL AFFAIRS COMMITTEE, AMERICAN ASSOCIATION OF CLINICAL ANATOMISTS (1996). A Clinical Anatomy Curriculum for the Medical Students of the 21st Century: Gross Anatomy. *Clin Anat*, 9: 71-99.

FONTAINE CH (2001a). Some thoughts about anatomic variations. *Surg Radiol Anat*, 23: 1-2.

FONTAINE CH (2001b). Some help for literature study in anatomical variations reports. *Surg Radiol Anat*, 23: 293-294.

GRIFFIOEN FMM, DRUKKER J, HOOGLAND PVJM and GODSCHALK M (1999). General plan anatomy, objectives of the teaching of the anatomy/embryology in medical curricula in the Netherlands. *Eur J Morphol*, 37: 288-325.

HAST MH and GARRISON DH (2000). Vesalius on the variability of the human skull: book I of the Humani corporis fabrica. *Clin Anat*, 13: 311-320.

HOLMES LB (1976). Congenital malformations. *N Eng J Med*, 295: 204-207.

MOORE KL (1989). Meanning of "normal". *Clin Anat*, 2: 235-239.

JONES DG, DIAS GJ, MERCER S, ZHANG M and NICHOLSON HD (2002). Clinical anatomy research in a research-driven anatomy department. *Clin Anat*, 15: 228-232.

LIPPERT H and PABST R (1985). *Arterial variations in man, classification and frequency*. J.F. Bergmann-Verlag, München.

MARDEN PM, SMITH DW and MCDONALD MJ (1964). Congenital anomalies in the newborn infant, including minor variants. *J Paediatrics*, 64: 357-371.

MÉHES K and STADLER G (1983). *Minor malformations in the neonate*. Akadémiai Kiadó, Budapest.

SINGER CH (1956). *Galen on anatomical procedures*. Oxford University Press, New York, Oxford.

SINGER CH (1957). *A short history of Anatomy and Physiology from the Greeks to Harvey*. Dover Publications, New York.

STEVENSON RE and HALL JG (1993). Terminology. Vol I. In: Stevenson RE, Hall JG and Goodman RM (eds). *Human malformations and related anomalies*. Oxford University Press, London, pp 21-30.

STRAUS WL and TEMKIM O (1943). Vesalius and the problem of variability. *Bull History Medical*, 14: 609-633.

VESALIUS A (1542). De Humani Corpori Fabrica (Ed. Spanish, 1997). Editorial Ebrisa, Madrid.

WILLIAM PLT and HUMPHERSON JR (1999). Concepts of variation and normality in morphology: import issues at risk of neglect in modern undergraduate medical courses. *Clin Anat*, 12: 186-190.