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FETAL COMPRESSION AND THE RECOGNITION OF CONGENITAL DEFORMATION, 1960-1981 *

Peter M. Dunn,

MA, MD, FRCP, FRCOG, FRCPCH

Emeritus Professor of Perinatal Medicine and Child Health

University of Bristol

(P.M.Dunn@bristol.ac.uk)

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In August 1967 I was invited to speak at a perinatal conference organised by the University of Florida and held at the Plaza Hotel at Daytona Beach, home of many famous car races. I was delighted to find that the distinguished perinatal pathologist, Edith Potter, was also on the faculty[1] (Fig.1).

Fig.1 Dr. Edith Potter (1901-1993)

Born in 1901, she qualified in medicine at the University of Minneapolis in 1925. Nine years later in 1934, she was appointed first as instructor and then as professor of pathology at the Chicago Lying in Hospital. In 1952, on the basis of some 10,000 necropsies, she published her great work entitled 'The Pathology of the Fetus and Infant'[(2)]. Almost single handed Potter founded the new modern specialty of perinatal pathology. She died in 1993 at the age of 92.

Since her death in 1993, Edith Potter has been best remembered eponymously by the facial characteristics of infants born with bilateral renal agenesis[(3)]. She first described this facies in 1946 when she wrote:

> 'The peculiar facies of these infants seems to have no specific embryologic correlation with the renal anomaly. The face most characteristically exhibits an increased space between the eyes, a prominent fold which arises at the inner canthus and sweeps downward and laterally below the eyes, an unusual flattening of the nose, with excessive retraction of the chin ... and a moderate enlargement and decreased chondrification of the ears. The face gives a suggestion of premature senility and is sufficiently characteristic to warrant a diagnosis of co-existing renal aplasia when it is observed.' (Fig.2)

Edith Potter also observed that these babies usually had limb deformities such a club feet. In addition she recorded that they invariably had pulmonary hypoplasia . However, she was unable to explain the presence of this hypoplasia, pointing out that: 'There is no apparent relationship between

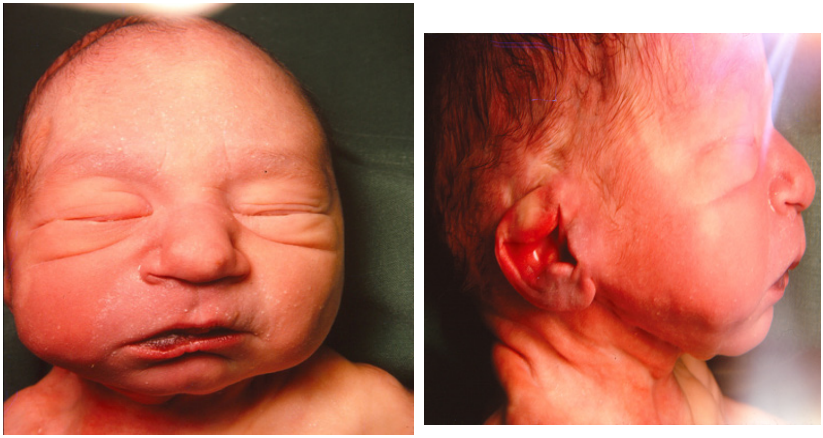

Fig.2a and 2b Potter's facies (front and side views).

the embryonic development of the lungs and the ureters and kidneys'. Although she recorded the probable association of the lack of amniotic fluid with bilateral renal agenesis, she did not relate the characteristic facies and associated deformities with prenatal fetal compression.

I remain especially grateful to Edith Potter in that the sight of a case of Potter's Syndrome in 1958 helped to stimulate my interest in prenatal fetal deformation. During the decade 1959-1968 I worked on a magnum opus entitled: *'The influence of the intrauterine environment on the causation of congenital postural deformities with special reference to congenital dislocation of the hip'*[(4)]. When after three years of clinical research I began to study the literature on this subject, I found to my surprise that the notion that intrauterine constraint might lead to fetal deformation, though discounted in recent times, actually reached back to the days of Hippocrates[(5)]. It had never been discussed when I was a medical student and there

was no mention of the idea in any of my medical textbooks. Furthermore I discovered that the idea had been vigorously advanced by Sir Denis Browne[6] (Fig.3) in 1934, though without achieving wide support. The orthopaedic world in particular dismissed and derided the idea that mechanical forces *inutero* might lead to fetal deformity.

The problem with the research of Sir Denis and others was

Fig.3 Sir Denis Browne (1892-1967)

that it was purely observational, unsupported by convincing scientific evidence.

However, my own observations were supported by such evidence in the form of epidemiological studies on a cohort of 6,756 consecutively newborn infants studied between 1960 and 1963. This enabled me to confirm my conclusions with the aid of 180 statistical analyses, some of them to be seen in Figure 4, which reveals that the main congenital postural deformities occur in association with each other.

CONGENITAL POSTURAL DEFORMITIES	Facial def.	Plagioceph.	Mandib. asym.	Sternom. contr.	Scoliosis	C.D.H.	Talipes
Facial Deformities		S	S*	S	S*	S*	S*
Plagiocephaly	S		S*	S*	S*	S*	N
Mandibular asymmetry	S*	S*		S*	N	S*	S*
Sternomastoid contr.	S	S*	S*		S*	N	S*
Scoliosis - postural	S*	S*	N	S*		S*	S
Cong. Disloc. Hips	S*	S*	S*	N	S*		S*
Talipes	S*	N	S*	S*	S	S*	

N = not signif; S = P<0.05; S* = P<0.001

Fig.4 Statistical analysis of studies made during 1960-63 of the clinical association between certain congenital postural deformities (Dunn 1969(4)). Abbreviations: N: not significant; S: P<0.05; S*: P<0.001

Besides clinical and radiological studies I also made many pathological observations on this subject, courtesy of two pathologists, Dr. Hans Kohler in Birmingham, and Dr. Norman Brown in Bristol. All together I must have undertaken four or five hundred post-mortem examinations.

Returning to the subject of Potter's facies, perhaps the most fundamental observation I made in the early 1960s was of the highly significant association of Potters facies and other deformations with maternal oligohydramnios, whether due to bilateral renal malformation, whatever its nature – agenesis, polycystic kidneys, etc.-, or to prolonged leakage of amniotic fluid, or to the oligohydramnios associated with severe placental insufficiency. It was also highly significant that Potter's facies and limb deformities were <u>not</u> present when renal malformation was unilateral. However, they <u>were</u> present when there was severe urinary tract obstruction below the level of the bladder causing oligohydramnios. The critical factor was, of course, whether or not the fetus was able to pass urine *inutero*, and hence provide volume to the amniotic fluid.

Let me give two brief case histories in support of these observations:

> Case 1: Maternal oligohydramnios. Breech delivery at 37 weeks gestation. Male infant weighing 3.4Kg with Potter's facies and respiratory distress. Bilateral pneumothoraces drained. Died at 18 hours. Necropsy revealed urethral valves and dilation deformities of the bladder, ureters and kidneys. Pulmonary hypoplasia was present.

Case 2: Maternal oligohydramnios secondary to premature rupture of the membranes and prolonged drainage of the amniotic fluid. Male infant with Potter's facies and postural deformities of the hands and feet (Fig.5). Kidneys and urinary tract normal.

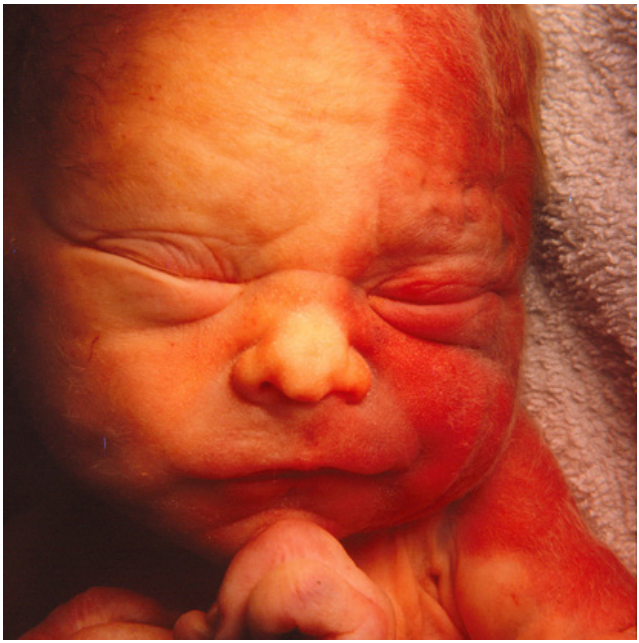

Fig. 5 Infant with classic Potter's facies and postural deformities secondary to oligohydramnios due to leakage of amniotic fluid. Kidneys and urinary tract normal.

In May 1968 my three volume thesis [4] was submitted to the University of Cambridge. Eight months were to pass before I was informed that it had been finally accepted for the degree of MD. Later, I heard that one of the assessors,

a professor of orthopaedic surgery, had given it the thumbs down. Fortunately, the other assessor and an extra one called in to adjudicate had eventually given it their approval. Many papers based on my thesis followed[7-15]) and in 1976 I was asked to summarise my findings in an edition of the British Medical Bulletin devoted to 'malformations'[16].

The distinguished Professor Tom McKeown of Birmingham, an expert in the etiology and epidemiology of congenital anomalies, was invited to review this edition of the bulletin. He commented: 'The significance of mechanical influences in the uterus on congenital postural deformities (reviewed by Dunn) is still an open question.'[17] Fortunately, not everyone was sceptical. In 1975 I was invited to take part in an International Conference on the Classification of Congenital Anomalies[18] at the National Institute of Health, Bethesda, under the Chairmanship of Dr. David Smith, author of the classic text, 'Recognisable Patterns of Human Malformation'[19]. Both at that meeting and at a subsequent conference on the same subject in Baltimore chaired by Dr. Victor McKusick, I presented my ideas on the origin of congenital postural deformities. To my delight, they again received very strong support.

But what was my thesis?[4,16] In summary, it was that: 'Quite gentle forces, if persistently applied, might lead to deformation. That such deformation occurs much more readily in the presence of growth. That the fetus is particularly vulnerable to deformation because of its rapid rate of growth and relative plasticity. That prenatal deforming forces might be intrinsic or extrinsic in origin. That most fetuses were exposed to

extrinsic forces in the later weeks of pregnancy because of their increasing size and the diminishing volume of amniotic fluid. That at least 2% of infants exhibited postural deformities at birth though the great majority of these deformities either resolved spontaneously or responded to early postural correction. Figure 6 lists most of the main anomalies that I have termed the congential postural deformities in relation to the part of the body affected.

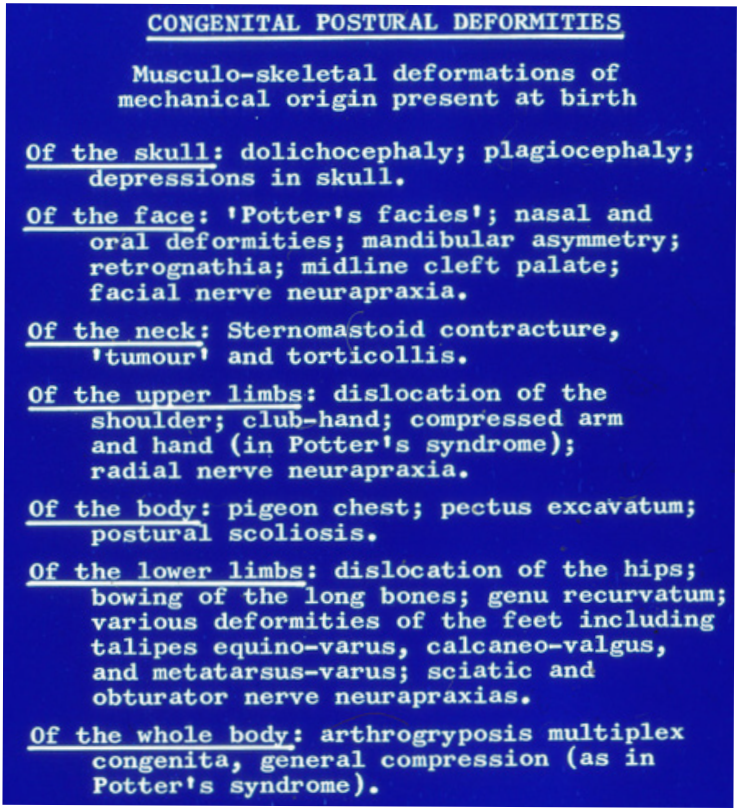

Fig.6 Congenital postural deformities in relation to the part of the body affected.

With the aid of epidemiological statistical analyses, it was possible to show that the various deformities seen in the last figure not only occurred in association with each other to a highly significant degree (Fig.4) but also occurred in association with the following pregnancy factors – first pregnancy, breech presentation, oligohydramnious, maternal hypertension and fetal growth retardation. Moreover, it was also possible to demonstrate how these pregnancy factors were related to each other and to the causation of fetal deformation[4,16] (Figs. 7 and 8).

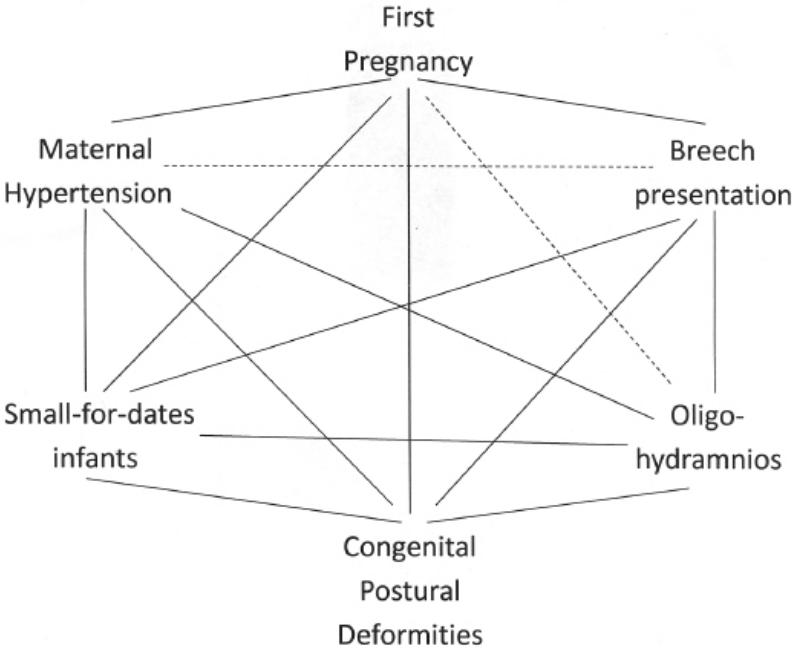

Fig. 7 Congenital postural deformation and certain pregnancy factors. (Dunn 1969[4]). Each unbroken line represents a statistically significant association, while the interrupted lines represent probable but unproved associations

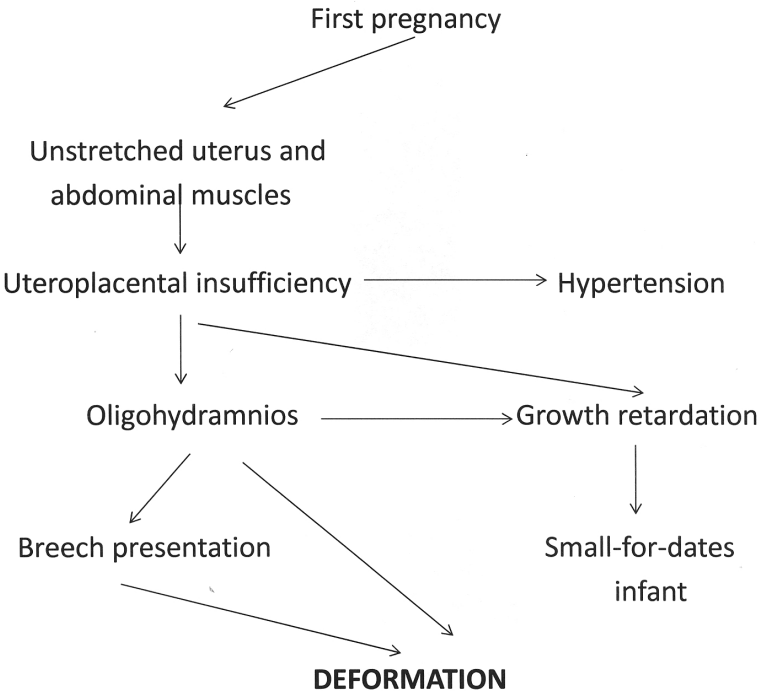

Fig. 8 Possible interrelation of some of the pregnancy associations with congenital postural deformation (Dunn 1969[4]).

The underlying significance of my thesis was that while malformations were defects arising during the period of organogenesis and were essentially teratological embyropathies, congenital deformations were defects arising after the embryonic period and were thus alterations in a previously normally formed part of the body. They were fetopathies. The importance of this distinction is absolutely fundamental

to the understanding of their occurrence and management, as well as to the prognosis of these deformed infants involving some 2% of all newborn babies.

The support I received at those two conferences in the USA in 1975 was particularly helpful as at that time I had been engaged by the World Health Organisation to help revise the 1967 8th edition of the International Classification of Diseases[20]. In chapter 14 of this revision, dealing with congenital anomalies, what I personally termed the postural deformations were all mixed up together with the malformations and were only related in the text to the anatomical part of the body involved. While it is not easy to bring about changes in the ICD, which is only revised about once a decade, I am happy to say that in ICD 9 (1979) my arguments in support of the separation of deformities from malformations were accepted with the result that the deformities were then segregated from the malformations with their own anatomic rubrics[21]. As you may imagine, it gave me great satisfaction after having been told for many years that my ideas were ill-conceived. Later in the 1980s I was also invited to help to prepare the 10th revision of the International Classification of Diseases[22]. This provided another opportunity to further improve and bed down the deformation rubrics.

But there was yet another pleasure in store. In 1977 David Smith, doyen of dysmorphologists, rang me up and asked if he and his wife could come and stay with us in Bristol (Fig.9). It transpired that he had come over from Seattle, in order to read and photocopy my MD thesis[4] held in the Cambridge University Library.

Four years later in 1981 David Smith's secretary sent me, at his request, a copy of his new textbook, a companion to his earlier one, with the title: 'Recognisable Patterns of Human Deformation'[23].

Fig.9 Prof. David Smith of Seattle with Mrs. Anne Smith (1977).

Typical of the man, he had paid me generous credit in the preface. He wrote:

> 'It was in February, 1975, at an international meeting on Nomenclature for Birth Defects, that I first heard Peter Dunn, MD, of Bristol, England, give his impassioned plea for a clear distinction between defects due to mechanical

constraint forces (deformation) as contrasted to those due to poor formation (malformation). His recommendation was accepted forthwith. Since that time, I have been ever more impressed by the importance, relevance, and magnitude of this deformational category of birth defects. Hence, I acknowledge Peter Dunn as the individual who set me forth on the pathway that culminated in this book.'

I would, of course, have liked to have been able to thank David Smith both for the book and his kind acknowledgement but, sadly, I learnt that he had died one month before its publication

I fear as Shakespeare once said, 'All this comes too near the praising of myself'. Please forgive me but over the whole 50 years since I first formulated my thesis on the nature of congenital postural deformation, this tribute by David Smith was the only public acknowledgement I have ever received. What had been considered a misconceived idea, overnight had become an obvious truth. Meanwhile the author, previously considered to be a crank, has become a bore. Such is life!

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