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Prenatal diagnosis: The irresistible rise of the 'visible fetus'

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ABSTRACT

Prenatal diagnosis was developed in the 1970s, a result of a partly contingent coming together of three medical innovations—amniocentesis, the study of human chromosomes and obstetrical ultrasound—with a social innovation, the decriminalization of abortion. Initially this diagnostic approach was proposed only to women at high risk of fetal malformations. Later, however, the supervision of the fetus was extended to all pregnant women. The latter step was strongly favoured by professionals' aspiration to prevent the birth of children with Down syndrome, an inborn condition perceived as a source of suffering for families and a burden on public purse. Experts who promoted screening for 'Down risk' assumed that the majority of women who carry a Down fetus will decide to terminate the pregnancy, and will provide a private solution to a public health problem. The generalization of screening for Down risk increased in turn the frequency of diagnoses of other, confirmed or potential fetal pathologies, and of dilemmas linked with such diagnoses. Debates on such dilemmas are usually limited to professionals. The transformation of prenatal diagnosis into a routine medical technology was, to a great extent, an invisible revolution.

1. Introduction: the 'PND dispositif'

Prenatal diagnosis (PND), or rather the 'PND dispositif'—a dynamic and constantly evolving array of techniques and approaches which provide information on the fetus during pregnancy—has radically changed the experience of pregnancy for tens of millions of women. From the 1940s on, gynecologists took a growing interest in the fetus, and developed strategies to assess fetal health (Arney, 1982; Casper, 1998, pp. 30–72). These efforts culminated in the 1970s with the development of the 'PND dispositif'. This dispositif came into being thanks to a partly contingent coming together of three medical technologies—amniocentesis, the study of human chromosomes and obstetrical ultrasound—with a social innovation, the decriminalisation of abortion. At first, prenatal diagnosis was proposed only to women classified as being at high risk of fetal malformations. Later, however, in many industrialized countries, screening for fetal malformation was extended to all pregnant women. Pregnancy itself became a 'risk factor'.

Gynaecologists always aspired to promote the birth of healthy children, and reduce the danger of premature birth, childbirth complications and perinatal mortality. However, until the 1980s, they focused on the management of pregnant women's bodies (Al-Gailani, 2010). Feminist critique of an excessive medicalisation of pregnancy was directed against doctors' attempts to regulate the behaviour of pregnant women (Oakley, 1984), By contrast, in the 21st century, the supervision of pregnancy is to a significant degree centred on the fetus, as are the anxieties of pregnant women. This radical change of perspective, this paper argues, was to an important extent the result of professionals' aspiration to prevent the birth of children with Down syndrome, a congenital condition redefined in late 20th century as an important public health problem. The generalization of testing for 'Down syndrome risk' favoured in turn the intensification of screening for other fetal conditions. While the supervision of the bodies and lifestyles of pregnant women continues to be an important part of prenatal care, from the 1980s on the fetus became increasingly the main focus of medical and lay attention.[1]

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[1] There is, nevertheless, significant variability in attitudes to pregnancy in industrialized countries. Tsipy Ivry argues that in Japan pregnant women, and not the fetus, continue to be the main target of medical supervision (Ivry, 2007).

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2. From the amniotic tap to amniocentesis

The first element that made prenatal diagnosis possible was the development of a technique to sample the amniotic fluid. In the late 19th century, gynaecologists employed newly developed hollow hypodermic needles to siphon an excess of amniotic fluid, a condition called polyhydramniosis. Severe polyhydramniosis can be risky for the fetus and, in especially drastic cases, for the mother too. Polyhydramniosis is often (but not always) associated with diabetes. Pregnancy in diabetic women was very rare in the early 20th century, but became less exceptional, when, with the development of insulin, more women with this condition survived to adulthood and decided to have children. As a result, the treatment of polyhydramniosis—which, in the meantime, acquired a name, 'amniotic tap'—became less rare too.

Another reason for increased use of the 'amniotic tap' was the development of a test for presence of bilirubin in amniotic fluid. The yellow colour of the fluid, measurable in a spectrophotometer, was a sign of severe haemolytic disease of the fetus, a condition in which a Rh (Rhesus factor) negative mother produces antibodies against Rh positive red blood cells of the fetus. When haemolysis was detected, physicians often decided to induce birth, and/or perform an exchange transfusion immediately after the baby's birth (Bevis, 1952). Their first aim was to save the newborn's life. An additional and important goal was the reduction of the risk of damage produced by free bilirubin (kernicterus) and its neuropsychiatric consequences, such as deafness and mental retardation (Vogel, 1953; Worssam, 1957). Doctors affirmed that an amniotic tap allowed them accurately to predict the presence of a haemolytic disease of the newborn in 95% of cases (Walker, 1957). At first promoters of this method denied that the amniotic tap may be harmful for the fetus or the mother. In the 1960s, practitioners recognized however that this intervention, renamed amniocentesis, could lead to a miscarriage, and explained that it should be used only if considered absolutely necessary (Walker, Fairweather, & Jones, 1964).

Amniotic fluid contains cells shed by fetal membranes. Its sampling thus provided access to fetal cells. The first such studies investigated fetal sex. In 1948 a Canadian anatomist, Murray Barr, observed the presence of a marker, the Barr body, in female but not male cells (Miller, 2006). By the mid-1950s, several research groups that analyzed aborted fetuses had shown that it is possible to determine fetal sex through the study of cells in the amniotic fluid. Early recognition of fetal sex has practical importance for the diagnosis of sex-linked hereditary diseases. In 1960, two Danish researchers, Povl Riis and Fritz Fuchs, analyzed cells in the amniotic fluid of a pregnant woman who was a carrier of haemophilia (haemophilia is X-chromosome linked disease; women are carriers but do not get sick; half of their male children will have the disease). They showed that the fetus was female, and the woman subsequently gave birth to a healthy girl (Riis & Fuchs, 1960).

At that time, a Danish act of 1956 made 'eugenic' abortion legal. The interruption of pregnancy for 'conductors of hereditary diseases' was granted by a board of the Danish Mothers' Aid Institution. Riis's and Fuchs's first patient knew that she was a carrier of the haemophilia gene. She had already had one legal abortion and decided to go ahead with her next pregnancy but the child, a boy, haemorrhaged and died hours after his birth. When she found out that she was pregnant again, she decided to terminate the pregnancy, then was informed about the possibility of finding out the sex of the fetus. By contrast, Riis's and Fuchs's second patient learned about her risk of giving birth to a child with haemophilia only when she was already four months pregnant. Amniocentesis revealed again that the fetus was female, but the woman miscarried. Riis and Fuchs did not indicate whether she was informed about the risk of this procedure (Riis & Fuchs, 1960; Cowan 2008). In the 1960s, Riis and Fuchs performed 16 additional prenatal diagnoses of fetal sex in haemophilia carriers. In every case but one, a diagnosis of male sex was followed by abortion. One woman, affected by an 'emotional factor' (the death of her father), changed her mind and decided to continue the pregnancy. One diagnosis of male sex was erroneous; after the fetus was aborted, Riis and Fuchs discovered that it was female (Harris, 1971, pp. 141–145).

The Danish law, which allowed an abortion for a hereditary risk of severe and non-curable abnormality or physical disease, was unmistakably eugenic, not only in name but also in content (Paul, 1995, pp. 114–115). In the early 1960s, historical links between abortion and eugenics were, however, weakened by two consecutive environmental disasters that put at risk all pregnant women, not only those from 'tainted' families.

3. Thalidomide, rubella and the changing view of abortion

In the late 1960s and 1970s, abortion for 'maternal distress' became legal in the majority of Western countries. This legislative shift is often linked to the 'sexual revolution' of the 1960s, the rise of post-1968 feminism and changes in women's status.[2] However, these elements did not lead directly to the decriminalization of abortion. In the second half of the twentieth century, women in many countries—Ireland, Poland, and nearly all Latin American countries—achieved greater sexual freedom, amplified their access to education and the professions, and increased their participation in economic and political life (today, some of these countries are led by female politicians), but are still unable legally to terminate a pregnancy. Inversely, the legalization of abortion in Japan in the 1950s was not related to feminist struggles, greater sexual freedom, or women's increased participation in the work force. Changes in sexual mores and in women's status undoubtedly favoured the modification of abortion laws in Western Europe and North America. It is plausible to assume, however, that two disasters: the thalidomide scandal of 1961–62, and the German measles epidemic of 1962–64, also had an important effect on attitudes towards the termination of pregnancy.

In 1961, a widely diffused tranquilizer, thalidomide, was found to be responsible for the birth of deformed children on a massive scale. The drug was popular among pregnant women because it alleviated morning sickness in early pregnancy. In 1961, an Australian physician, William McBride, published a letter to the *Lancet*, linking the drug's uptake by pregnant women to the appearance of severe birth defects in children, especially the absence of limbs (phocomelia) (McBride, 1961). Other researchers confirmed these findings (Leck & Millar, 1962; Rodan, Koller, & Taylor, 1962; Ward, 1962). A famous American paediatric surgeon, Helen Brooke Taussig, had heard about the suspicion that thalidomide induced fetal and neonatal malformations from a German colleague, and went to Germany find out more. Taussig became persuaded that thalidomide was indeed responsible for the observed malformations, but only if taken at a precise moment during early pregnancy. The narrow time window in which thalidomide could harm the fetus explained why only a fraction of the pregnant women who took this drug gave birth to abnormal babies. Taussig wrote a paper about the German epidemics of fetal malformations for *Scientific American*, and insisted that it should be illustrated with

[2] The 'sexual revolution' is often explained by the development of the contraceptive pill. Liberalization of sexual behaviour, however, preceded, the development of reliable contraceptive means (Löwy, 2010; Smith, 1990).

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