

"Screening for Congenital Hypothyroidism" (1991), by Delbert A. Fisher ^[1]

By: Craer, Jennifer R. Keywords: fetal iodine deficiency disorder ^[2] endemic cretinism ^[3]

In his 1991 article "Screening for Congenital Hypothyroidism," Delbert A. Fisher in the US reported on the implementation and impact of mass neonatal screening programs for congenital hypothyroidism (CH) from the early 1970s through 1991. CH is a condition that causes stunted mental and physical development in newborns unless treatment begins within the first three months of the newborn's life. In the early 1970s, regions in Canada and the US had implemented screening programs to diagnose and treat CH as quickly as possible after the infant's birth. By 1991 many other countries had adopted the early screening program, and Fisher estimated that 10 to 12 million newborns per year were tested in the early 1990s. The screening programs, along with physician education and improved screening techniques, such as radioimmunoassay, helped significantly reduce the incidence of abnormal newborn development resulting from untreated congenital hypothyroidism.

Fisher specialized in the study of children's [hormones](#) ^[4], and he worked as a physician and researcher at the University of California, Los Angeles medical school in Torrance, California. He published "Screening for Congenital Hypothyroidism" in *Trends in Endocrinology & Metabolism* in 1991. The article has four sections. The first section provides background information about CH and the screening programs. The second section describes neonatal thyroid disorders. The third section offers a prognosis for affected infants, and the fourth section demonstrates the role that screening programs have played in combating CH.

In the first section, Fisher describes congenital hypothyroidism as a condition recognized for nearly two centuries as a major cause for impaired growth and mental development. Fisher highlights the work in the early 1970s of Salvatore Raiti and George H. Newns in England, and of Alan H. Klein in the US, who showed that infants with CH develop more like infants without CH when doctors diagnose and treat CH before afflicted infants are three months old.

Fisher also stresses that the radioimmunoassay (RIA) is useful for early detection of CH. Rosalyn Yalow and Solomon Berson in the 1950s in New York, New York developed RIA, a technique that uses radioactive isotopes as markers to measure minute concentrations of biochemical substances from small blood samples. Fisher notes the expanded applications of RIA, which measure iodothyronines and thyroid-stimulating [hormone](#) ^[5] (TSH) in blood levels. Iodothyronines and TSH contribute to the development of thyroid [hormones](#) ^[4], and TSH stimulates the thyroid to produce thyroid [hormones](#) ^[4]. If doctors are to identify and treat thyroid disorders, Fisher says, then they need to recognize abnormal levels of iodothyronines and TSH. Fisher touts the low cost, requirements for small samples of blood, and sensitivity of RIA as factors that led to successful neonatal screening programs for CH.

Fisher continues in the first section of "Screening for Congenital Hypothyroidism," by highlighting neonatal screening programs for mass populations around the world. In 1974

Jean Dussault and colleagues introduced the first mass-screening program for CH in Quebec, Canada. By the end of the decade, additional sites in North America began screening programs. Parts of Western Europe, as well as Japan and Australia, quickly adopted the screening programs. The programs screened twenty-five million infants by 1982. By 1991 countries throughout Eastern Europe, South America, Asia, and Africa had also adopted screening programs, making it possible for ten to twelve million infants to be screened per year.

Fisher concludes the first section by describing how RIA methods enhanced CH screening. In 1974, Dussault and colleagues included the newly developed thyroxine or T4, (one of the thyroid [hormones](#) [4],) RIA assay with the ongoing phenylketonuria (PKU) screening program to utilize its existing infrastructure and reduce costs for CH screening. In 1975 Reed Larsen and Kathy Broskin in Pittsburgh, Pennsylvania modified Dussault's method to more rapidly measure T4 concentrations in blood samples. Further developments included a more sensitive TSH assay. Some screening programs use a combined T4-TSH assay, or a T4 assay alone followed by a TSH if T4 concentrations are lower than normal. Elevated TSH concentrations point to abnormal thyroid function, but Fisher recommends that doctors screen neonates between 3 and 5 days old to avoid the normal surge of TSH [hormone](#) [5] seen in neonates shortly after birth.

In the second section of "Screening for Congenital Hypothyroidism," Fisher describes a continuum of neonatal thyroid dysfunction. He lists seven causes of thyroid dysfunction, of which three are transient disorders, treatable and temporary conditions. If detected and treated within three to five days of birth, physicians have ameliorated or prevented the stunted development of the neonates. The first disorder is hypothyroxinemia. Transient hypothyroxinemia is prevalent in premature infants and results from temporary insufficient production from the hypothalamus of thyrotropin releasing [hormone](#) [5] (TRH), a [hormone](#) [5] needed to stimulate the production of TSH. In parts of the world where people lack iodine in their diets, drugs or antibody-induced blockages of the TSH receptors cause the second transient disorder, primary hypothyroidism. In contrast, the third disorder, transient hyperthyrotropinemia, is caused in a [fetus](#) [6] *in utero* when the pregnant woman is overexposed to iodine.

Fisher next outlines the four causes of permanent thyroid dysfunctions in fetuses and neonates. Permanent disorders occur in roughly one in 4,000 births. Abnormal development of fetuses' or neonates' thyroid glands, called sporadic thyroid dysgenesis, accounts for eighty-five to ninety percent of permanent cases. Abnormal production of thyroid [hormones](#) [4], or thyroid dyshormonogenesis, accounts for between five and ten percent of the permanent disorders and leads the thyroid gland to swell. A third condition, hypothalamic-pituitary-hypothyroidism, is caused when a neonate's hypothalamus produces too little thyrotropin releasing [hormone](#) [5] (TRH), a [hormone](#) [5] that helps produce TSH. Hypothalamic-pituitary-hypothyroidism accounts for three to four percent of the permanent disorders. Diagnosis of the fourth condition, generalized resistance to thyroid [hormone](#) [5] (GRTH), usually does not occur within the neonatal period.

The third section of "Screening for Congenital Hypothyroidism" discusses the prognosis of infants born with hypothyroidism. Fisher contends that screening programs have improved the health of infants diagnosed and treated early. Fisher reports that neonates treated for CH have similar IQ scores, physical growth, and bone maturation compared to non-CH afflicted children when they are six to eight years old. Unsuccessful cases of treatment involved

children who may have had delayed treatment or inadequate T4 treatment. Fisher says that early detection and appropriate treatments are imperative to the future health of an affected infant.

Fisher assesses the effectiveness of screening programs in the final section of "Screening for Congenital Hypothyroidism" and highlights a 1985 screening program in the Northwest US. The program initially screened 812,000 infants, and when it retested 485,000 of the infants up to six weeks later, it found that fourteen infants had elevated TSH levels not present at the earlier test. Fisher then compares these findings with a similar report from the New England Congenital Hypothyroidism Collaborative published in 1979, which found, from a collective 1.2 million screened, eight infants demonstrated the same rise in TSH. Based on the combination of these records, Fisher estimates that one in 100,000 infants could be at risk of delayed elevation of TSH levels. Fisher devotes the remainder of the fourth section to promote the need for clinical detection of CH in developing infants. Fisher offers a list of symptoms to watch for in newborns: slow growth, slow weight gain, delayed developmental milestones, poor feeding, and constipation. Other symptoms include [jaundice](#) [7], enlarged tongue, umbilical hernias, and an enlarged posterior fontanelle. Fisher encourages healthcare providers to refer to neonatal screening records and to test for current [hormone](#) [5] levels.

At the conclusion of the fourth section, Fisher reviews the history of neonatal screening and its role in reducing common side effects of CH in affected infants. Since the employment of neonatal screening programs, more than twelve million infants were screened annually in the industrialized world. Despite the screening programs, Fisher asserts that doctors fail to diagnose 12 percent of infants with CH, and he urges doctors to be alert for the possibility of undiagnosed CH.

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Subject

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