Letters to the Editor

Neonatal Screening for Congenital Metabolic and Endocrine Disorders

Results From Germany for the Years 2006–2018

by Dr. med. dent. Anja Lüders MPH, Dr. med. Oliver Blankenstein, Dr. med. Inken Brockow, Prof. Dr. med. Regina Ensenauer, PD Dr. med. Martin Lindner, Prof. Dr. med. Andreas Schulze, and Dr. med. Uta Nennstiel MPH in issue 7/2021

Support Deficit in Adulthood

Thank you very much for the study (1), according to which a lot has been achieved; however, there is still a lot to be done. An early detection program in routine care can only be ethically justified if all subsequent steps after early detection are organized so that better care is provided for persons with a newly discovered illness. Due to the early detection, children become adults requiring lifelong treatment. There are around 1000 new cases per year, and hence an increased number of affected children are now becoming adults, 17 years after the introduction of screening in 2004.

Already 20 years ago, we (2) warned that the care of adults with these diseases, which can only be treated in a specialized way, must be systematically regulated, and that the nationwide introduction of special outpatient clinics is required. In addition, payment for this care must be available. Otherwise, an early detection that leads to nothing would be tolerated.

In 2002, there were five university hospitals that had outpatient departments for adults with these conditions. In 2014, there were only ten (3). Today, according to the Working Group for Congenital Metabolic Disorders (ASIM, *Arbeitsgemeinschaft für Angeborene Stoffwechselstörungen*), there are 17 outpatient clinics (4) throughout Germany that care for adults who did not exist 30 years ago, because children with congenital metabolic and hormonal disorders mostly died young.

In addition, there is still the problem of remuneration: the statutory health insurance (GKV) does not consider itself responsible for the consequences of early detection (apart from local special agreements or the much too low general polyclinic flat rate). So far, the state has also not considered itself responsible—although the program was introduced by it. ASIM (4) recently asked the Federal Ministry of Health to include adults as well as children in the draft law to improve care for this group of patients.

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Conflict of interest statement
The author declares that no conflict of interest exists.

Use Existing Registry

We read the article with great interest (1). We agree with the authors that newborn screening is a major achievement in pediatrics for the early diagnosis of serious diseases. We can only agree with the authors' request to establish a registry and continuous quality assurance. However, we miss the information that a registry for tracking patients has existed for the two endocrine diseases (congenital adrenal hyperplasia (CAH), in German *adrenogenitales Syndrom* [AGS], and congenital hypothyroidism) for over ten years (initially the AQUAPE registry; renamed by the German Society of Paediatric Endocrinology and Diabetes [*Deutsche Gesellschaft für Kinderendokrinologie und -diabetologie*] as the DGKED-Register, DGKED-QS).

The participating institutions (CAH (AGS): 53; hypothyroidism: 66) document indicators for a guideline-compliant therapy and course with standardized software and transmit anonymized data twice a year for evaluation. From this, benchmarking reports are created for each institution, and the results are published together (2, 3).

The registry currently includes 30 687 examination appointments for 1773 patients with CAH (AGS), and 22 504 examination appointments for 1920 patients with congenital hypothyroidism.

All pediatric endocrinology institutions in the German-speaking area are invited to participate in these registries (contact: www.peda-qs.de) in order to come closer to the goal of the most comprehensive possible follow-up of the patients recorded in the newborn screening.

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The authors declare that no conflict of interest exists.

In Reply:

We agree with Prof. Abholz's demand for adequate, competent care also for adult persons with rare diseases discovered in newborn screening. In this context, the necessity of a structured transition for an organized transfer from pediatric and adolescent medicine to adult medicine, and the recognition of possible obstacles, should be pointed out (1–3).

We are happy to support the invitation made by Prof. Holl and Prof. Wölfle to all endocrinology institutions to participate in the DGKED registry. We had previously discussed presenting the registry; however, a complete recording of the cases of the disease seems to be impossible under the current legal conditions, especially for hypothyroidism. In our experience, more than 60% of children with congenital hypothyroidism are treated by a local pediatrician. This situation should have been discussed in the article, which unfortunately was not possible due to lack of space.

As already mentioned in the article (4), we consider it to be absolutely necessary to comprehensive record all persons with diseases discovered during screening in registry structures, for quality assurance as well as to evaluate the success of newborn screening. As the incomplete coverage rate of existing registries shows, both data protection issues for the secure registration of all persons and the effort involved in data entry must be more clearly regulated in order to dissolve the existing dependency on the commitment of the institutions involved.

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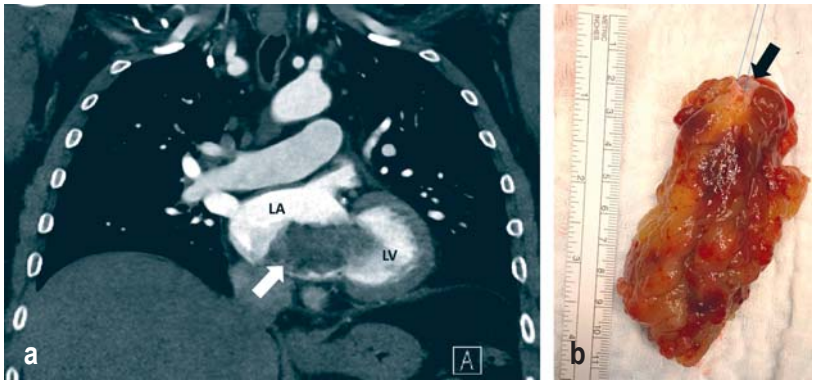
Dr. Nennstiel has received reimbursement of travel expenses and lecture fees from Biogen.

CLINICAL SNAPSHOT

Giant Left Atrial Myxoma and Left Ventricular Inflow Obstruction



a) Coronal section of a computed tomography angiography of the heart with contrast medium in the left ventricle and left atrium. The black filling defect of contrast medium (arrow) indicates the mass.
b) Macroscopic specimen of the surgically excised myxoma (size: 9 × 5 × 5 cm). The arrow marks the "endocardial button."

A 58-year-old patient developed dyspnea after undergoing a total hip replacement. Computed tomography revealed an intracardiac mass (arrow in *Figure a*). Transesophageal echocardiography was performed as part of further diagnostics. This showed an inhomogeneous structure originating in the left atrium and extending to the left ventricle. The 9 × 5 × 5-cm structure with its 1 × 1-cm "endocardial button" (endocardial attachment site—arrow in in *Figure b*) was successfully excised using an atrial transseptal approach under cardiopulmonary bypass. Histopathology confirmed the diagnosis of myxoma. The postoperative course was unremarkable and no recurrence was seen at 1-year postoperative follow-up. Myxoma is the most common primary cardiac tumor; it is benign, generally located in the left atrium, and usually smaller than in this case. Compared to cardiac thrombi, myxomas tend to be larger, are polypoid in shape, and are mobile structures due to the "stalk" that is usually present. The recurrence rate following myxoma excision is under 5%.

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