What is microcephaly?
The term microcephaly is based on the Greek words ‘mikros’ and ‘kephal’ and means ‘small head’. Microcephaly is a condition in which the head is significantly smaller than that of other people of the same age, sex and ethnic background.
The head (skull) consists of bones, which embrace and protect the brain. In the first years of life, until brain growth is completed, the head circumference adapts to the extension of the brain. Thus, the head grows with the brain. This is why microcephaly can occur in any disorder that affects brain growth.
In rare cases, microcephaly can result from a closure of the growth plates of the skull before the brain has stopped growing. This so-called craniosyostosis is associated with skull deformity as the brain tries to push outward in alternative directions and can impair brain development.
How is microcephaly diagnosed?
Microcephaly is diagnosed by measuring the head circumference with a non-elastic measuring tape. The head circumference measurement is evaluated using standard curves and tables. Microcephaly is present if the measured head circumference is under the third centile or if it lies more than two standard deviations below the mean for age, sex and ethnicity.

How is microcephaly classified?
Microcephaly can be classified as primary or secondary type. Microcephaly is considered to be primary if it is already present at birth, opposed to secondary microcephaly which develops later in life.

Is microcephaly a disease?
Microcephaly refers to a clinical sign rather than a disease. It may or may not be associated with disease, i.e., the presence of microcephaly does not automatically imply illness. However, microcephaly is a risk factor for neurologic (and often rare) diseases and for other conditions that affect health and quality of life. Therefore, your physician may advise further examinations after a detailed discussion with you and a physical examination of your child. Such tests may include urine and blood analysis, imaging (e.g. ultrasound, MRI scans) and consultations with other specialists. The prognosis of a child with microcephaly depends on the etiology, the extent of microcephaly and associated malformations.

How is the cause of microcephaly determined?
There are various causes of microcephaly. It can, therefore, be quite a challenge to determine the underlying disease amongst the abundance of possible conditions. Your physician will need to collect various data. This includes medically relevant information on your child and your family, physical examinations, laboratory tests, imaging and specialized examinations of organs such as heart, kidneys, eyes, ears and brain. There are guidelines for a standardized approach to patients with microcephaly. If initial testing does not lead to an accurate diagnosis, medical experts should be involved. In case microcephaly is suspected during pregnancy, prenatal examinations can be offered.

Who can I turn to?
Your pediatrician is the right person to first contact for all questions regarding microcephaly. The pediatrician may recommend to consult further medical specialists such as a pediatric neurologist and/or a geneticist. For questions concerning prenatal diagnostics please contact your gynecologist.

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