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Variety and variation in sex development – support individuality

Sometimes it is not possible to determine at birth whether the child is a boy or girl. But a rethink by medical science in recent years means that irreversible surgery can now be avoided as far as possible. Variation in sex development and rare childhood hormone disorders will be the focus of discussions at the annual conference of the European Society for Paediatric Endocrinology (ESPE) at the Austria Center Vienna from 19-21 September.

“As paediatric endocrinologists we specialise in diagnosing and treating very rare illnesses in children,” explained Professor Gabriele Häusler, Head of the Department of Pediatrics and Adolescent Medicine at the Medical University of Vienna and President of ESPE 2019. “Which is why we have chosen variety and variation as the theme for this year’s ESPE Congress. We want to allay some of the fears surrounding rare phenomena such as variation in sex development and send out a signal promoting tolerance towards statistical deviations from the ‘norm’,” Häusler explained.

Variation in sex development – everyone is an individual

“Every person is an individual,” stressed Dr. Stefan Riedl, a paediatric endocrinologist at Vienna’s St. Anna Children’s Hospital and Vice President of ESPE. In many cases, variations in sex development manifest at birth, but on some occasions they only emerge during puberty, when a boy starts to develop breasts or a girl does not start menstruation. With some variations of sex development it is clear which sex the child should be assigned to – as with a boy whose urethra ends on the underside of the penis instead of at the tip of the glans. This particular condition affects around one in every 300 boys. But sometimes the definitions of “boy” and “girl” are too narrowly defined – these profound variations are, as Dr. Riedl confirmed, extremely rare, with a prevalence of around 1 in 4,000. The causes of variations in sex development are as diverse as the conditions themselves and can be chromosomal, genetic, physiological or endocrinological – i.e. hormonal – in character.

Variation in sex no longer has to be a barrier to having children

Insufficient production of cortisol by the adrenal glands can indirectly lead to the external genitalia of a girl resembling those of a boy at birth. Hormone replacement therapy in the first week following birth is necessary for survival, and the sex variation is a symptom that can lead to the clitoris looking like a penis as a result of imbalanced testosterone production. An insufficiency of the same hormone in boys can lead to the external genitalia not taking on a typically male appearance. “With some variations in sex development, identity is not contested and it will be possible for the individual to reproduce when they reach the age of sexual maturity,” Riedl confirmed. In the case of genetic forms of variations in sex development,

infertility is probable in adulthood – but even here fertility can potentially be ensured, for example, in XXY individuals healthy sperm can be extracted from the testicle in puberty for use later.

Greater caution instead of irreversible treatment

Variations in sex development is a highly sensitive topic – particularly when they do not fit the black and white male/female mould. “In previous generations, there was a strong tendency to operate at a very early age and shroud things in a veil of silence,” explained Riedl. “But we are seeing a change in the way that people think today. Children are seen as individuals, families receive psychological support and self-help groups form part of the management process,” he said. “Only specific cases are treated early – in addition to medically necessary hormone therapy, these include lengthening the urethra or removing non-functional gonads which could develop into tumours. In all other cases as much time is allowed to pass as possible, until puberty or later, in line with the principle of bodily integrity,” the experienced endocrinologist added. If an individual approaching puberty is not sure of their sexual identity, hormone blockers provide a way of delaying the onset of sexual maturity.

Paediatric endocrinology – specialising in rare disorders

“In addition to variations in sex development, as paediatric endocrinologists we work with paediatricians to diagnose suspected hormone disorders such as growth and thyroid disorders. Our work also extends to the field of genetic bone disease and manifest skeletal growth disorders – such as brittle bone disease, achondroplasia and hypophosphatemic rickets,” said Häusler, highlighting the scope of this far-ranging specialism.

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