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URGENT DIAGNOSIS IN PRACTICAL GENETIC COUNSELLING

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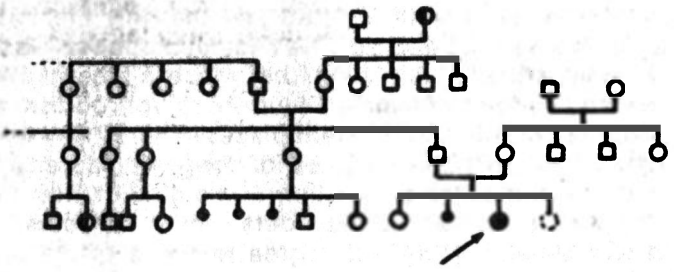
Urgent diagnosis in practical genetic counselling has to be done most often in case of anomalies in the sex differentiation which makes difficult the determination of the gender of the newborns and their timely notification in the civil registry of the country. The work of the geneticist in "urgent" examination of the genetic gender in newborns with inborn genital malformations is less known. Having that in mind, we discuss those cases in the last 3 years of the Genetic Counselling Center, Varna Medical University in which "urgent" diagnosis of the genetic sex has been done.

Genetic sex was examined in 7 newborns of the town and the district of Varna by cytogenetic methods because of various anomalies in the differentiation of the genitals. Female gender was specified in 5 of the babies and male one - in 2. In 3 cases with female gender genital anomalies were isolated and the only manifestation was virilization in a various degree. The clinical diagnosis was "Congenital adrenal hyperplasia" (virilizing form which is inherited AR). All the patients with this form of female pseudohermaphroditism are minority and their families live in different places. That is why we could suggest high frequecy of heterozygotes for congenital adrenal hyperplasia among this part of the population of the district. This point of view was supported by the pedigree data where we found "clitormegalia" in two females, II and III degree relatives of the proband (fig.1). The sex anomalies of the other two babies with female gender were part of nonsystematic multiple malformations. Both families had previous reproductive failures but the corresponding pregnancy and the delivery has been normal. The babies had normal weight and hight at birth. In the first case the inborn anomalies were with unclear etiology and nosology which includes clitormegalia with hypoplasia of labia minores. The cytogenetic study did not show any numeral and structural aberrations of the normal female karyotype 46,XX. This case, in our opinion, needs further specifying of the clinical diagnosis and prognosis of the later physical and psychological development of the child as well as finding the reasons for this condition.

The second proband is a baby with uncertain gender, omphalocele, bladder extrophy, open symphisis, fusion of vagina and anus.

Post mortem cystic fibrosis was also proved. Different data show very low recurrence risk for bladder extrophy, but in this case it is combined with AD inherited defect (omphalocele) and AR disease (cystic fibrosis). Because the couple has not got alive child the genetic investigation must include preliminary DNA analysis for typization of the cystic fibrosis gene while the next pregnancy must be checked for all defects. Genetically determined male gender we proved in 2 babies with genital anomalies. The first one was born after four years sterility. The mother became pregnant a year after ending the sterility treatment, no data for professional or other noxious factors. The newborn was with low weight (1800 g) and hight (42 cm). Genital anomalies included: micropenis, hypospadia, doubled scrotum, palpation of the testicles (not very certain), hymen. The necessity of later determination of the gonadal gender and the possibilities of better correction of the condition were discussed in the genetic counselling. The second baby with cytogenetic male gender had very young minority parents - 17-year old mother and 20-year old father who were close relatives. The couple had before this child one healthy girl.



● clitormegalia ● defect in external genital organ formation

Fig.1 A pedigree of congenital adrenal hyperplasia (AR) patient

In conclusion we would like to point out that 4 of the newborns who needed "urgent" diagnosis of the genetic gender were minority, 1 was from a family of Bulgarian moslems. These couples usually have lower social position and/or live in isolated populations which makes difficult the well-timed and adequate correction of the defects as well as preventing anomalies of the genitals in their off-springs. On the other hand, these difficulties do not repeal the necessity of accurate and precise genetic counselling and collaboration between the geneticist and different clinical specialists.