Reasons for disuse preimplantation genetic diagnosis in carriers with familial amyloidotic polyneuropathy

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**Background:** Familial amyloidotic polyneuropathy (FAP) is a hereditary autosomal dominant disease, whose largest focus in the world is Northern Portugal. In 2001, preimplantation genetic diagnosis (PGD) was introduced as a way to identify the Met30 mutation in the Portuguese-type FAP (type I). This study aims to estimate the prevalence of FAP carriers who have undergone PGD, and to detail reasons surrounding their disuse of such technique.

**Methods:** Between January and April 2013, a representative sample of 195 Portuguese FAP type I carriers, aged between 18 and 55 years and followed at the national surveillance unit, was systematically recruited. Data on sociodemographic, reproductive and obstetric history, and reasons underlying the use of PGD were collected through a self-administered questionnaire. The current analysis is based on 79 patients with FAP trying to get pregnant between 2002 and 2013.

**Results:** The prevalence of PGD use was 19.0% (95% CI: 11.0 to 29.4). The main reason for disuse PGD (n=64) was unknown the technique (29.0%). Additionally, one quarter (25.8%) reported technical distrust (misdiagnosis or undiagnosis, lower success rates, and child adverse outcomes), followed by financial costs (24.2%) and lengthy period of time to get pregnant (9.7%). All those who reported unknown PGD had at least one children born after January 2002 (p=0.041). FAP carriers with less than 35 years (82.5% vs. 71.8%, p=0.257), more than 9 years of education (83.3% vs. 82.4%), and
72.1%, p=0.236) and household income above 2000€/month reported more frequently to known PGD (87.5% vs. 69.6%, p=0.064).

**Conclusions:** Less than 20% of FAP type I carriers who tried to get pregnant between 2002 and 2013 had undergone PGD. Main reasons justifying the disuse included lack of knowledge, distrust, and financial costs. These motives reinforce the importance of cognitive dimensions at the expense of psychosocial factors. This study highlight the need of incorporating accurate information into healthcare to improve informed decision making processes surrounding PGD use among these patients, focusing on the poorer, older, and less educated. Furthermore, a broaden access to PGD in the case of FAP type I carriers may contribute to improve the use of this technique.

**Main messages:**

High disuse of PGD among Portuguese FAP carriers (81.0%) was justified by lack of knowledge, distrust, and financial costs.

It's important to provide accurate information and to improve accessibility.