Why new diagnostic criteria for different PCOS phenotypes are urgently needed

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The Polycystic Ovary Syndrome (PCOS) is a heterogeneous disorder affecting between 5 and 15% of reproductive-aged women worldwide, depending on diagnostic criteria used. Today we recognize four phenotypes of PCOS, based on the presence or absence hyperandrogenism (HA; biochemical and/or clinical), ovulatory dysfunction (OA; reflected as poly-, oligo- or amenorrhea), and polycystic ovarian morphology (PCOM), after the exclusion of mimicking or similar disorders (thyroid dysfunction, hyperprolactinemia, congenital adrenal hyperplasia, etc.). These four phenotypes are ***Phenotype A***: HA+OA+PCOM (aka 'classic' or 'full' PCOS), ***Phenotype B***: HA+AO (aka 'classic PCOS'), ***Phenotype C***: HA+PCOM (aka 'ovulatory PCOS'), and ***Phenotype D***: OA+PCOM ('non-hyperandrogenic PCOS'). One way to distinguish different disorders is to assess their genetic underpinnings. To date it has not been possible to determine whether the genetic make-up of these four phenotypes differ, as most genetic studies have included small numbers of subjects with Phenotypes C and D, and because to date we have identified less than 10% of responsible genes. Another is to determine the heritability of the phenotypes in individual families. Although limited, current data suggests that individuals affected with phenotypes A, B & C may be present in the same family. Another approach is to determine the disorders' clinical consequences or long-term morbidity. In this case, the behavior of these phenotypes differs significantly. While Phenotypes A & B are closely associated with metabolic dysfunction and associated long-term morbidities, Phenotype C is less so, and Phenotype D almost not at all. A final confounder is the presence of concomitant obesity, whose prevalence varies widely worldwide and which appears to be relatively constant within affected individuals. Overall, further research into the origins and differences of the PCOS phenotypes is urgently needed.