OP157

TOWARDS A NATIONAL SCREENING PROGRAM FOR COELIAC DISEASE AND TYPE 1 DIABETES: AN ITALIAN FEASIBILITY PILOT PROJECT (D1CE SCREEN)

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Introduction: Celiac disease (CD) is an autoimmune disorder triggered by gluten ingestion in genetically predisposed individuals, leading to chronic intestinal inflammation. It affects about 1% of the global population and is often undiagnosed in childhood due to the absence of symptoms, with around 65% of cases in Italy going unnoticed.

Early screening is a key tool for timely diagnosis and preventing long-term complications. In this perspective, the D1Ce Screen pilot study follows Italian Republic Law 130/2023, which mandates screening for pre-symptomatic CD and type 1 diabetes (T1D) in the general paediatric population through autoantibody measurements in capillary blood. Paediatric screening is a key tool for early detection and the prevention of long-term complications, associated with late or missed diagnosis.

Individuals who tested positive for CD and T1D -specific autoantibodies will undergo further clinical evaluation at the regional specialized center to confirm the diagnosis

Aims & Methods: Primary objective of D1Ce Screen study is to assess, feasibility, acceptability and sustainability of a nationwide screening program, for the evaluation of undiagnosed CD cases and presymptomatic T1D. To assess operational aspects D1Ce Screen evaluates the involvement of primary care pediatricians (PCPs), blood sample collection accuracy and acceptability for families measuring anxiety, depression and quality of life through 3 validated questionnaires.

This is an observational multicenter study to screen 5,363 children in four Italian regions (Lombardy, Marche, Campania, Sardinia), distributed into three classes of age (2-2.9, 6-6.9, and 10-10.9 years), corresponding to peaks of seroconversion for these diseases. For eligibility, the inclusion criterion is age. T1D is the exclusion criterion. Voluntary PCPs were responsible for recruitment of participants, administration of informed consents and questionnaires, execution of blood drawing by finger prick, capillary blood collection and sample shipment.

Specific T1D and CD autoantibodies are measured by ELISA in capillary samples. In addition, HLA DQ2 and DQ8 HLA typing is performed on dried blood spot (DBS).

Results: PCPs initially accepting to participate were 565, with 428 (75,7%) completing the study. Enrolled children were 5534, with 91,1% within the target age. The gender distribution was 49.4% females, 50.6% males. Samples suitable for analysis (adequacy for at least one assay) were 89% of the total collected. Most of DBS samples were suitable for HLA typing. The target of families undergoing questionnaire evaluation of 2% has been exceeded.

Regions	Age (yr)							
	2+1	6+1	10+1	Out of range	In range	Total	Planned	%Planned
Campania	468	536	498	224	1502	1726	1739	86,37
Lombardy	824	936	1050	186	2810	2996	2867	98,01
Marche	113	123	127	19	363	382	394	92,13
Sardinia	96	126	147	61	369	430	363	101,65
Total	1501	1721	1822	490	5044	5534	5363	

Conclusion: The D1Ce Screen pilot study provides evidence supporting the feasibility of a nationwide screening initiative. The active collaboration of PCPs, the high level of participation from families, and the efficient implementation of capillary blood sampling procedures were key contributors to its success. These findings confirm the potential for scalability of a paediatric screening program for T1D and CD within the framework of the Italian National Health Service. Early detection and timely initiation of a gluten-free diet in children diagnosed with CD are essential to preventing long-term complications and improving health outcomes.

References:

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