

One in five children with diabetes and non-autoimmune extra-pancreatic features have monogenic aetiology

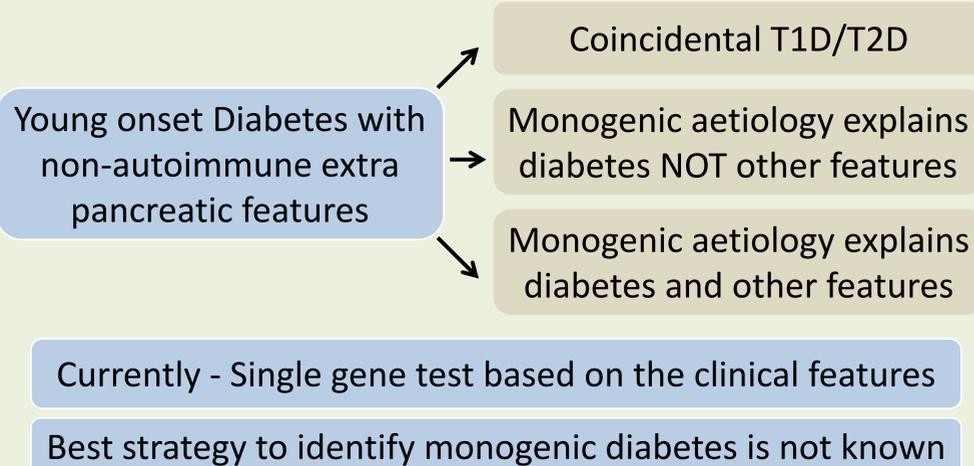


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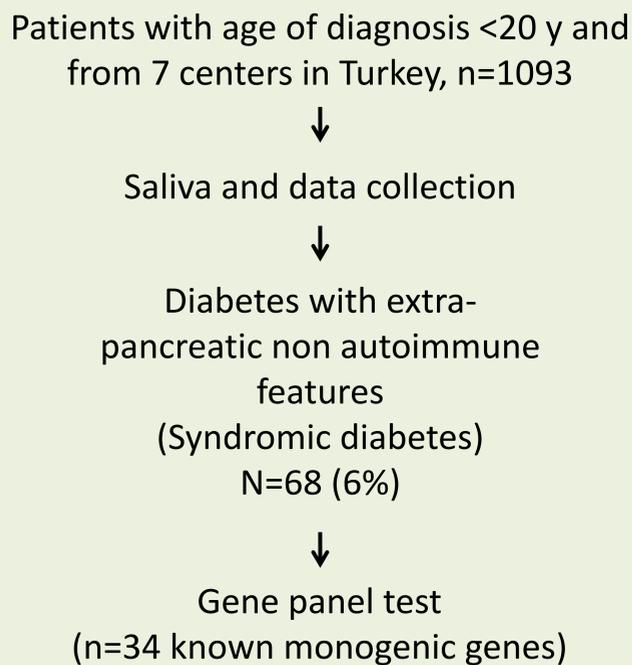
Background



Aim

Assess the utility of comprehensive genetic testing in children with diabetes and any non-autoimmune extra-pancreatic features.

Methods



Results

22% (15/68) children with syndromic diabetes had monogenic aetiology

Monogenic aetiology explains diabetes **NOT** other features
N=3 (20%)
GCK – 2, *PTF1A* p.Pro191Thr – 1

Monogenic aetiology explains diabetes **AND** other features
N=12 (80%)
WFS1 – 6, *SLC19A2* – 3, *SLC19A3* – 1, *TRM10A* – 1, *HNF1B* – 1

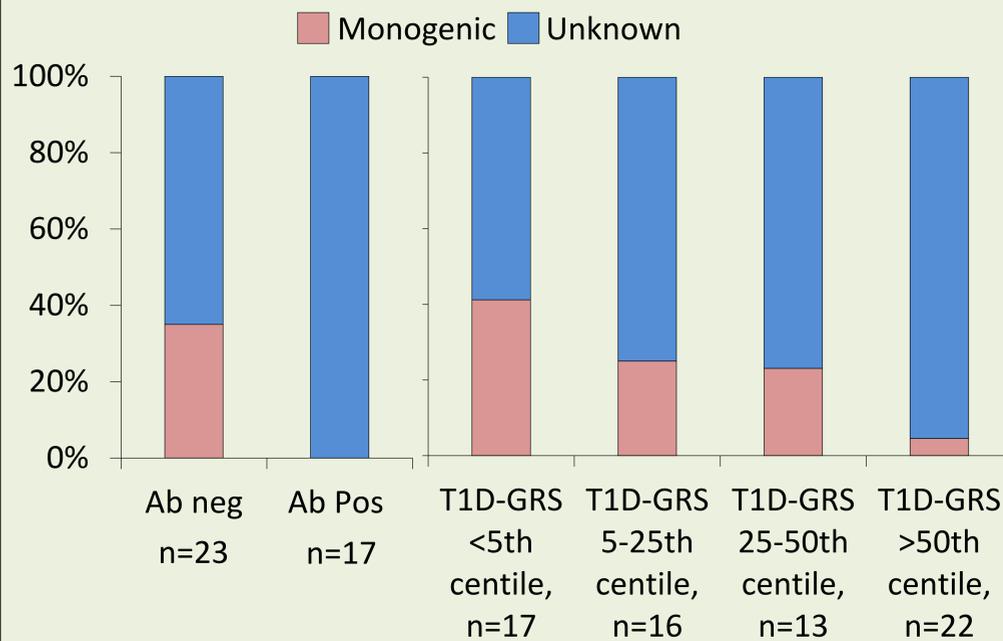
15% (2/12) had typical features indicative of underlying aetiologies

Results

Clinical features are similar between the unknown and known monogenic

	Syndromic Unknown, n=53	Syndromic Monogenic, n=15	P
Age of diagnosis (y)	8 (4.9-10.9)	6 (3.8-7.6)	0.2
Duration of diabetes (y)	3.3 (1.3-62)	4.7 (1.5-8.1)	0.5
BMI percentile	59 (30-87)	63 (24-91)	0.4
Female	28 (53%)	7 (47%)	0.7
Consanguineous parents	38 (72%)	6 (40%)	0.03
Parental diabetes	5 (9%)	2 (13%)	0.6
≥2 organs affected	13 (24%)	9 (60%)	0.01
Insulin Treated	49 (90%)	12 (80%)	0.3
Clinician reported Non-T1D	16 (30%)	6 (40%)	0.5

Islet autoantibodies and T1D-GRS help to identify monogenic diabetes



Conclusion

- 1 in 5 children with syndromic diabetes had monogenic aetiology.
- Gene panel test lead to early identification of syndromic forms of monogenic diabetes with minimal or atypical presentation.
- All children with diabetes and any non-autoimmune extra-pancreatic features should be considered for monogenic diabetes testing.
- T1D-GRS is a novel test that helps to identify children with high probability of monogenic aetiology.

