



Prenatal counseling and diagnosis of monogenic disorders: a two-center review of ten years in Hong Kong

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TABLE 1: BASELINE DEMOGRAPHICS

	N	%
NO. OF CASES (N)	64	100.0%
WIFE'S ATTENDANCE AGE (YEAR), MEDIAN, RANGE	33.9	(26.2 - 42.6)
AGE =< 35, N %	48	75.0%
AGE > 35, N %	16	25.0%
PREGNANCY ADULT, N %		
SPONTANEOUS	42	65.6%
PGT-M	22	34.4%
DISEASE HISTORY, N %		
CANCER PREDISPOSITION	11	17.2%
CARDIOVASCULAR	5	7.8%
NEURODEGENERATIVE	34	53.1%
RENAL	15	23.4%
DISEASE SYMPTOMATIC, N %		
HUSBAND SYMPTOMATIC	8	12.5%
WIFE SYMPTOMATIC	9	14.1%
BOTH COUPLE SYMPTOMATIC	0	0.0%
ASYMPTOMATIC	47	73.4%
PARITY (P) FOR CURRENT PREGNANCY, N %		
NULLIPAROUS	42	65.6%
MULTIPAROUS	22	34.4%
PREVIOUS HEALTHY PREGNANCY, N %		
YES	18	28.1%
NO	46	71.9%
MOLECULAR DIAGNOSIS, N %		
YES	44	68.8%
NO	20	31.3%
INFERTILITY, N %		
YES	15	23.4%
NO	49	76.6%
INVASIVE TESTING, N %		
YES	13	20.3%
NO	51	79.7%
PRENATAL INVASIVE DIAGNOSIS, N %		
AMNIOCENTESIS	8	12.5%
CVS	4	6.3%
NO DIAGNOSIS	52	81.3%
DELIVERY OUTCOME, N %		
HEALTH BABY	63	98.4%
ABORTION	1	1.6%

Design: A retrospective cohort study on our experience in prenatal counseling and management of pregnancies at risk of monogenic disorders in 2013- 2023 at Tsan Yuk Hospital (TYH) and Queen Mary Hospital (QMH) in Hong Kong.

Objective: To evaluate our experience in counseling and patient’s decision on prenatal diagnosis in monogenic disorders. We aim to compare patients’ decisions on prenatal diagnosis between pregnancies conceived naturally or via in-vitro fertilization- preimplantation genetic testing - monogenic defects (IVF-PGT-M).

Method: Amongst all couples referred during the period, couples at risk of monogenic disorders were identified. A retrospective review of medical records will be conducted on these individuals by analysing the written medical records and the electronic patient records.

Results: 64 cases were identified, and the median age of the patients was 33.9 years (range: 26.2–42.6). 42 cases (65.6%) were spontaneous conceptions, and 22 cases (34.4%) were IVF- PGT-M pregnancies. There was a similar distribution of patients’ ages between the natural and IVF-PGT-M groups (p = 1.00). There were no significant differences between the two groups in terms of disease symptoms, family history, gravida, parity, previous healthy pregnancy, and infertility. Statistically significant differences were noted in the disease categories, with a higher proportion of cancer predisposition in the PGT-M group (40.9% vs. 4.8%, p<0.001) and a higher proportion of renal disease in the non-PGT-M group (31.0% vs. 4.5%, p=0.023). Prenatal invasive diagnosis was done in 12 cases (18.8%). The IVF-PGT-M group had a higher rate of invasive testing (40.9% vs. 9.5%, p=0.0049) and a higher rate of molecular diagnosis and genetic testing (100% vs. 52.4%, p<0.001).

TABLE 2: COMPARISON OF NON PGT-M VERSUS PGT-M PREGNANCY ADULTS

	Overall	Non PGT-M	PGT-M		p-value ^a	-
NO. OF CASES, N %	64	42	65.6%	22	34.4%	
WIFE'S AGE OF ATTENDANCE, N %						1.00
AGE =< 35	48	31	73.8%	17	77.3%	
AGE > 35	16	11	26.2%	5	22.7%	
DISEASE CATEGORY ^B , N %						
CANCER PREDISPOSITION	11	2	4.8%	9	40.9%	<0.001
CARDIOVASCULAR	5	3	7.1%	2	9.1%	1.00
NEURODEGENERATIVE	34	24	57.1%	10	45.5%	0.44
RENAL	14	13	31.0%	1	4.5%	0.023
COUPLE SYMPTOMATIC ^B , N %						
HUSBAND SYMPTOMATIC	8	4	9.5%	4	18.2%	0.43
WIFE SYMPTOMATIC	9	6	14.3%	3	13.6%	1.00
ASYMPTOMATIC	47	32	76.2%	15	68.2%	0.56
BOTH COUPLE SYMPTOMATIC	0	0	0.0%	0	0.0%	1.00
FAMILY WITH AFFECTED, N %						0.80
MOTHER'S FAMILY	36	23	54.8%	13	59.1%	
FATHER'S FAMILY	28	19	45.2%	9	40.9%	
GRAVIDA (G) FOR CURRENT PREGNANCY, N %						1.00
G > 1	30	20	47.6%	10	45.5%	
G =< 1	34	22	52.4%	12	54.5%	
PARITY (P) FOR CURRENT PREGNANCY, N %						1.00
P > 0	22	14	33.3%	8	36.4%	
P = 0	42	28	66.7%	14	63.6%	
PREVIOUS HEALTHY PREGNANCY, N %						1.00
YES	18	12	28.6%	6	27.3%	
NO	46	30	71.4%	16	72.7%	
MOLECULAR DIAGNOSIS, N %						<0.001
YES	44	22	52.4%	22	100.0%	
NO	20	20	47.6%	0	0.0%	
GENETIC TEST, N %						<0.001
YES	43	21	50.0%	22	100.0%	
NO	21	21	50.0%	0	0.0%	
INVASIVE TESTING, N %						0.0049
YES	13	4	9.5%	9	40.9%	
NO	41	38	90.5%	13	59.1%	
INFERTILITY, N %						0.76
YES	15	9	21.4%	6	27.3%	
NO	51	31	73.8%	16	72.7%	

Conclusion: The method of conception (natural vs. IVF-PGT-M) influences patient decisions regarding prenatal invasive diagnosis.

This could inform future prenatal counseling approaches, emphasizing tailored counseling strategies based on the conception method and the disease category.

