什麼是*BoBs*®檢驗?

BACs-on-Beads®檢測方法 (下稱BoBs®) 能檢測到染色體13，18，21，X及Y數目異常及九種因微序列缺失而引起之遺傳性疾病。此檢測方法比傳統染色體核型分析更簡單，少量的樣本便足夠。同樣的樣本份量與*QF-PCR*比較，BoBs®能多測出九種容易被傳統染色體核型分析遺漏的疾病。在報告時間上BoBs®亦比傳統染色體核型分析及胎兒DNA檢測晶片為快。由於胎兒DNA檢測晶片會檢測出一些罕見的拷貝數變異令報告內容變得複雜，而BoBs®的報告結果相對直接精確。目前香港中文大學婦產科提供此項檢查服務。

所有需要做羊膜刺穿檢查或者絨毛膜取樣的孕婦都可作*BoBs*®檢驗

以下類別的孕婦特別值得做此項檢測：

1. 唐氏綜合症篩查屬高危
2. 超聲波檢查顯示胎兒有異常徵象
3. 家族史中曾有與BoBs®相關的遺傳性疾病

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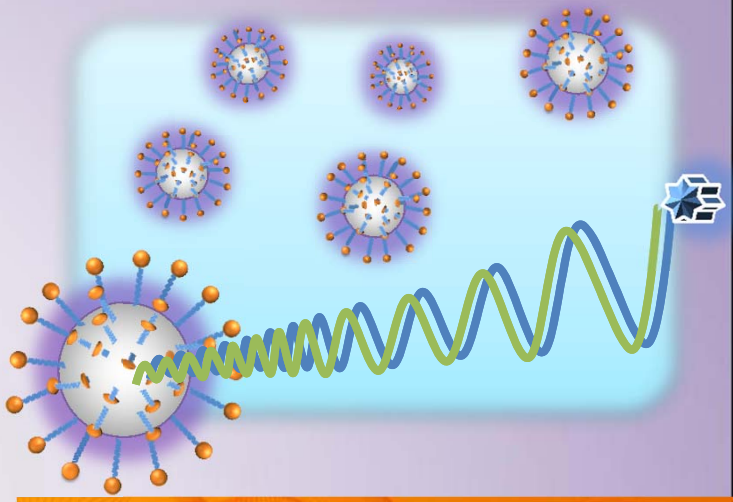

BoBs® *Assay*

不同檢測方法的比較

	傳統胎兒染色體測試		胎兒 DNA 檢測晶片	BoBs ®檢測
	染色體螢光定量快速測試	傳統染色體核型分析		
報告時間	1~2日	3周	7日內	2~3日
檢測涵蓋範圍	染色體21、18、13 、X及Y數目異常分析	染色體數目異常分析以及較大的染色體結構異常	染色體數目異常分析以及較大的染色體結構異常，超過一百種已知嚴重的基因疾病以及因微序列重複或缺失而引起之遺傳性疾病	染色體13, 18, 21, X及Y數目異常分析及九種遺傳性疾病(請見後頁表一)
價錢比較	$	$$	$$$	$
備註	不需要父母血液作分析		需要3-5ml父母血液作分析	不需要父母血液作分析

檢測樣本

1. 絨毛 *(11-14周)*
2. 羊胎水 *(16-21周)*
3. 血液 *(20周後)*
4. 胎盤
5. 妊娠組織

BoBs®檢測的限制

1. 報告結果不包括其他BoBs®沒有涵蓋的基因變異
2. BoBs®不能測出平衡易位、染色體倒位、點突變、染色體套數改變、單親二體症及低比例之鑲嵌性

What is *BoBs*® *Assay*?

BACs-on-Beads® Assay (BoBs®) is designed for rapid detection of common aneuploidy of chromosome 13, 18, 21, X & Y as well as nine known microdeletion syndromes. This simple and robust assay offers the advantage of ease of handling, high throughput and requires small sample volume. Compared with QF-PCR, *BoBs*® can detect 9 more microdeletion syndromes that might not be detected by karyotyping. Furthermore, the reporting time is greatly reduced when compared with karyotyping and Fetal DNA chip analysis. *BoBs*® can give a more straightforward and clear cut result as compared with Fetal DNA chip analysis which may detect some unknown Copy Number Variations (CNVs) and will complicate the result. The Department of Obstetrics and Gynaecology of the Chinese University of Hong Kong provides this new service.

Pregnant women undergoing invasive prenatal diagnosis may choose *BoBs*® *Assay*

Particularly for pregnant women with:

1. Positive Down's syndrome screening
2. Ultrasound findings of increase Nuchal Translucency (NT) or fetal structural abnormalities
3. Family history of genetic diseases covered by *BoBs*®

香港中文大學 婦產科學系

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表一: 可被*BoBs*®檢測出的三體綜合症及微缺失綜合症發生率
Table 1: Incidence rate of trisomies & microdeletion syndromes detected by *BoBs*®

病名	Disease Name	疾病發生率 (incidence rate)
沃夫-賀許宏氏症	Wolf-Hirschhorn Syndrome	1 in 50,000
貓鳴症候群	Cri-du-Chat Syndrome	1 in 20,000-50,000
威廉氏症候群	Williams-Beuren Syndrome	1 in 7,500
蘭-吉综合症	Langer-Giedion Syndrome	unknown
普瑞德威利症候群	Prader-Willi Syndrome	1 in 10,000-30,000
天使人症候群	Angelman Syndrome	1 in 12,000-20,000
無腦回畸形	Miller-Dieker Syndrome	1 in 25,000-85,470
史密斯-馬吉利氏症候群	Smith-Magenis Syndrome	1 in 15,000
顎心面综合症	DiGeorge Syndrome	1 in 6,395
巴陶氏症 (第13三體)	Patau Syndrome (Trisomy 13)	1 in 5,000
愛德華氏综合症 (第18三體)	Edwards Syndrome (Trisomy 18)	1 in 3,000
唐氏综合症 (第21三體)	Down Syndrome (Trisomy 21)	1 in 800-1,000
特纳氏综合症 (單體X性染色體)	Turner Syndrome	1 in 2,000-2,500

參考資料 (References):
GeneTests (http://www.ncbi.nlm.nih.gov/sites/GeneTests) and unpublished data

Comparison among different diagnostic methods

	Traditional chromosomal analysis		Fetal DNA Chip	*BoBs*®*Assay*
	QF-PCR	Karyotype Analysis		
Report Time	**1~2 days**	**3 weeks**	**Within 7 days**	**2~3 days**
Coverage	Aneuploidy of chromosome 13, 18, 21, X & Y	Aneuploidies and major structural chromosomal abnormalities	Aneuploidies and major structural chromosomal abnormalities Plus over 100 recognized genetic syndromes	Aneuploidy of chromosome 13, 18, 21, X & Y & 9 microdeletion syndromes (please refer to Table 1)
Price	**$**	**$$**	**$$$**	**$**
Remarks	**Couple's blood NOT required**		**3-5ml of couple's blood required**	**Couple's blood NOT required**

Samples required for this test

1. CVS *(week 11-14)*
2. AF *(week 16-21)*
3. Blood *(beyond week 20)*
4. Placental tissues
5. Product of Conception (POC)

Limitation of *BoBs*® *Assay*

1. Cannot detect gains and losses in chromosome regions not covered by *BoBs*®
2. *BoBs*® cannot detect chromosome inversions, balanced translocations, point mutations, ploidy changes, uniparental disomy, methylation alteration and low level mosaicism

若想知道更多資料，可向您的專科醫生查詢詳情，
或請瀏覽我們的網頁:
www.fetalmedicine.hk/en/oscarbooking.asp

For more details, please refer to your specialist
or visit our website:
www.fetalmedicine.hk/en/oscarbooking.asp

Please call us at (聯絡電話) 26326412