Down Syndrome

ROBERTSONIAN TRANSLOCATION

AN UNUSUAL PRESENTATION

Dr. Manvikar P R
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Dr.D.Y.Patil Medical College
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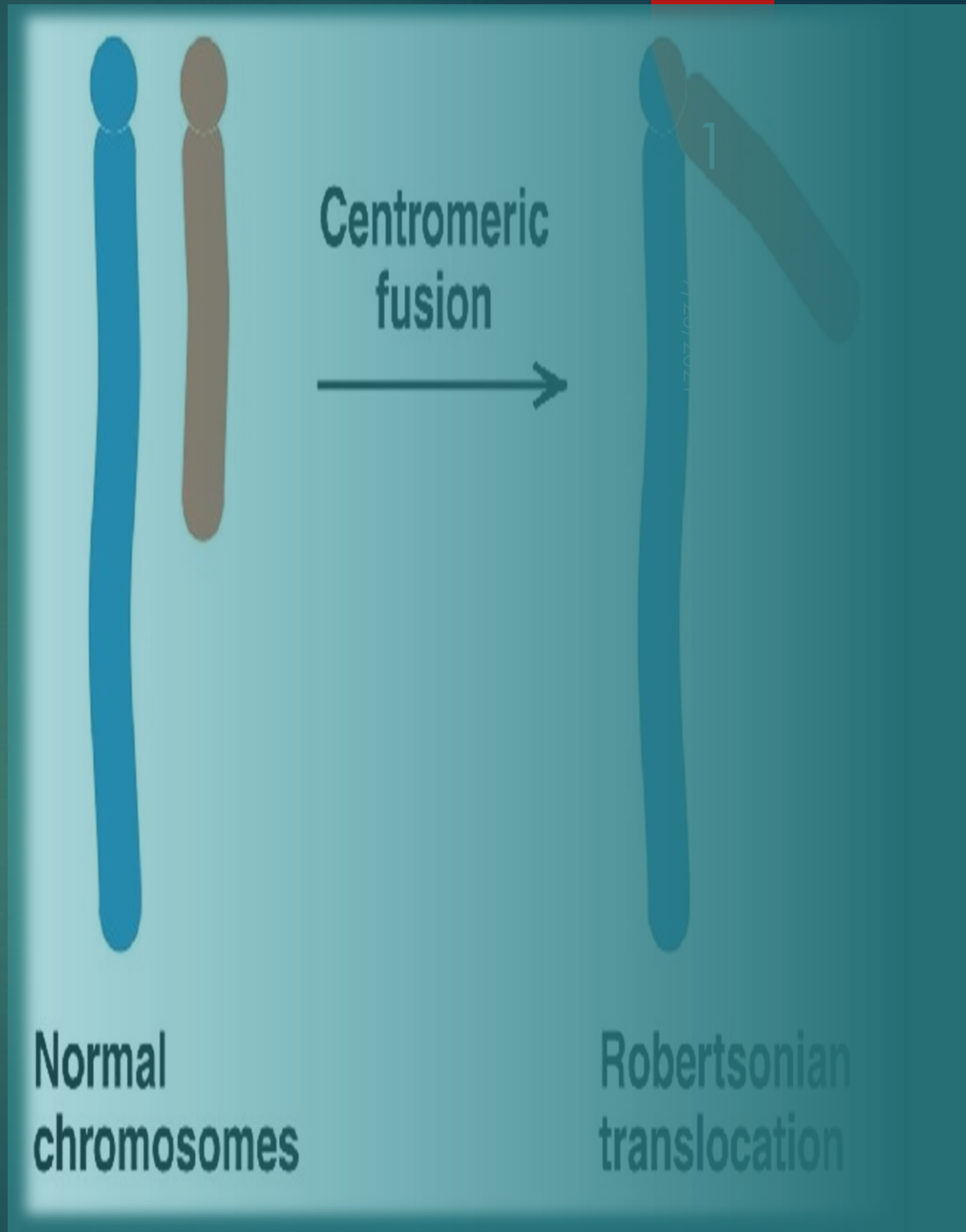


Case history

Proband :

- Male
- 8 months

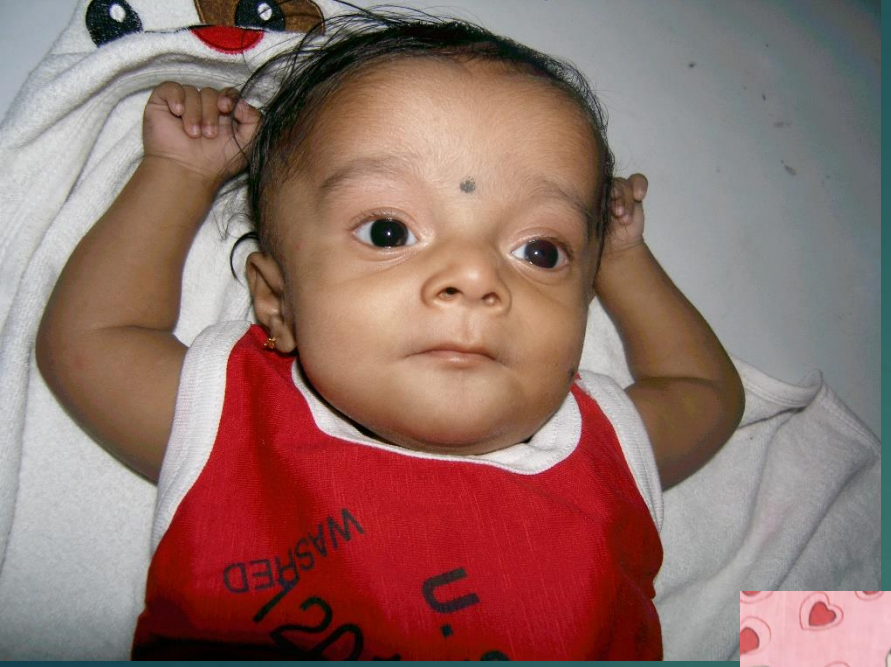
Family history

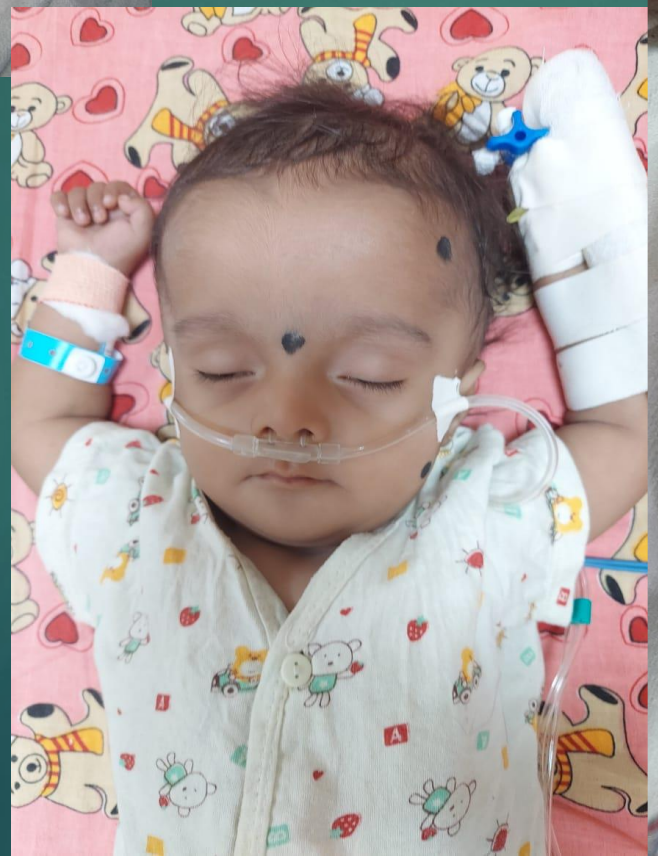
- Father
- 35yrs
- Bcom
- Marathi, hindu,
- Father h/o increased Bilirubin levels, non smoker, non tobacco user
- Non consanguinous marriage

- Mother :
- 25 yrs
- XII
- Marathi, hindu,
- Housewife
- Non consanguinous marriage

Clinical history

- Proband was referred for karyotyping by department of pediatrics
- History of URTI on admission
- Was found to be suffering from Facial dysmorphism, GDD, PDA, Hypospidias.
- H/O
  - triple marker negative for this pregnancy, FTND, Hospital delivery
  - Delayed milestones
- O/E
  - Esotropia
  - Low set ears
  - Increased space between Great toe and 2nd toe
  - Simian crease?(left)
- MRI: periventricular hyperintensities



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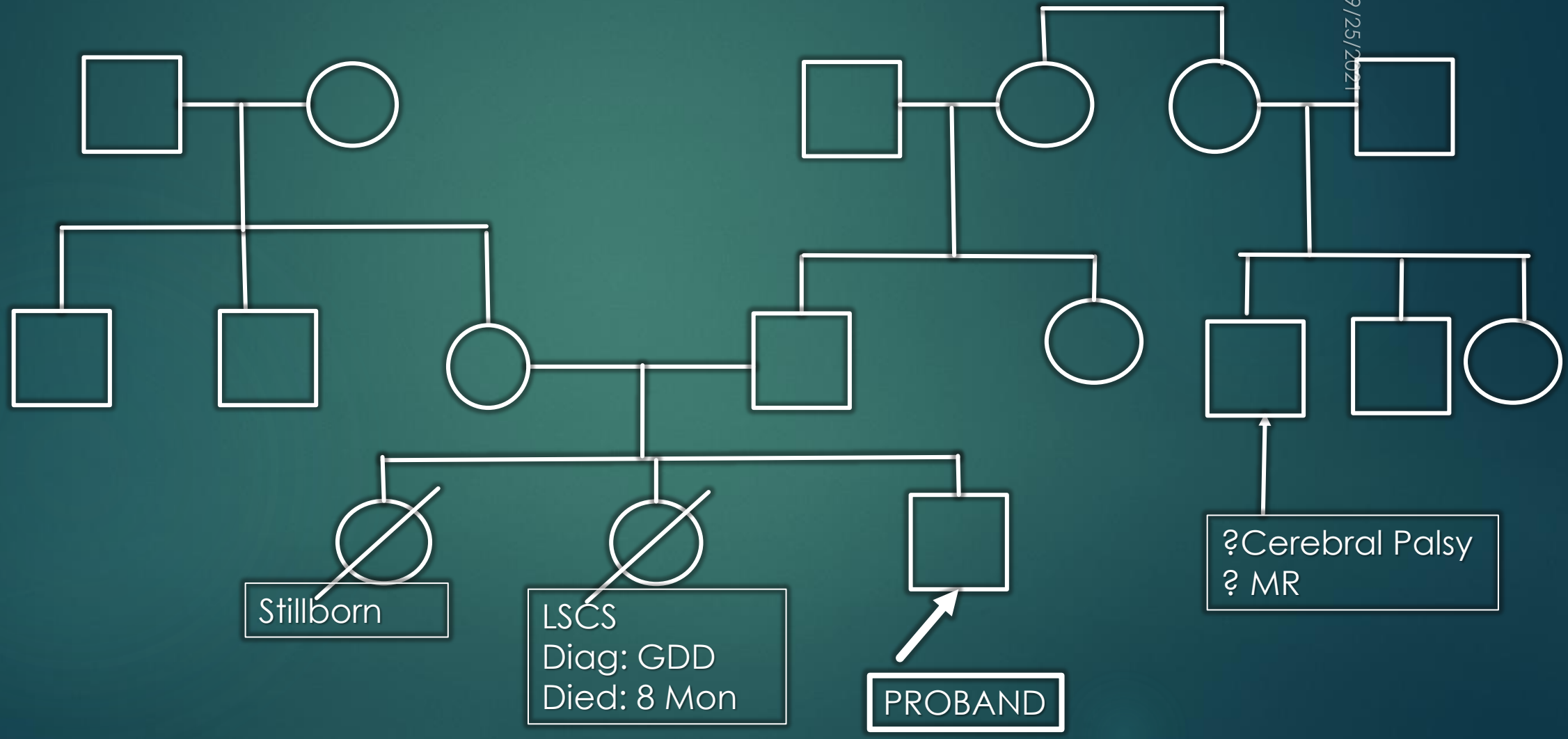
Differential Genetic Diagnoses

- Neiman pick
- Smith lemi optiz
- Sandoff
- Gangliosidosis

PEDIGREE CHART



BABY: PROBAND

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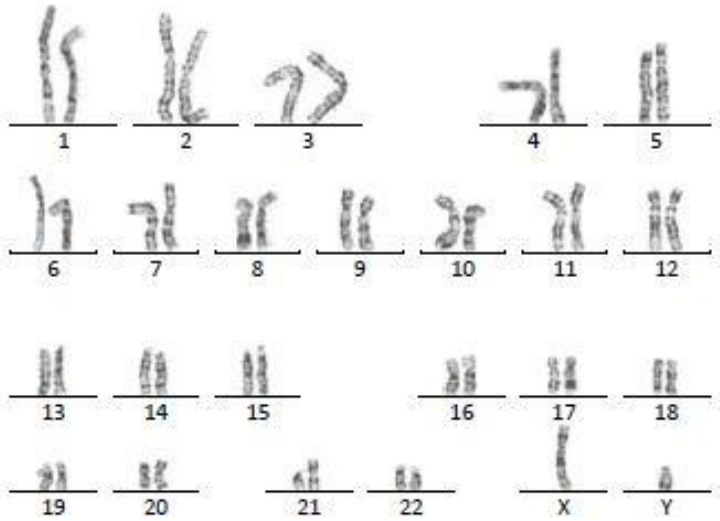
(Genetics Laboratory)

CHROMOSOME ANALYSIS REPORT

Patient ID: CG391 **Patient Name:** Satvik Patil **Age:** 8 month **Gender :** M
Physician Name / Hospital: Dr. S. Mane (Paeds)
Specimen: Peripheral Blood
Collected on: 22.03.2021 **Received on:** 22.03.2021 **Reported on:** 14.05.2021
Referral Reason: Global developmental delay, requested for karyotyping.



Banding: GTG

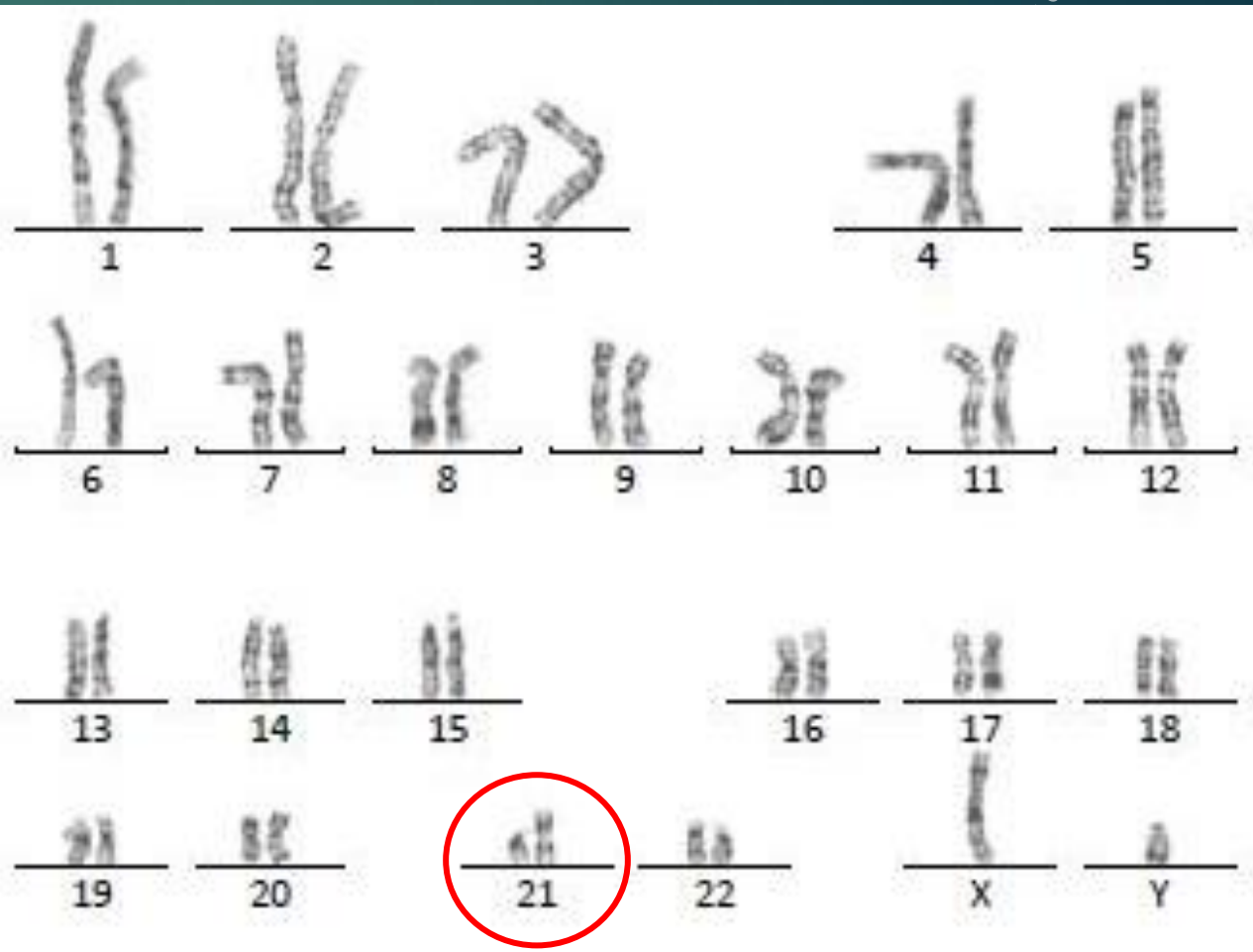
Karyotype: 46,XY,+21,rob(21;21)(q10;q10)
Co-ordinates : 99.1/4.6

Impression:
46,XY,+21,rob(21;21)(q10;q10)
Male karyotype with Trisomy 21
Suggested: FISH for chromosome 21

Note: Analysis is based on the sample received in the laboratory

Dr. Manvikar P.R.
(Cytogeneticist)

END OF REPORT



FATHER

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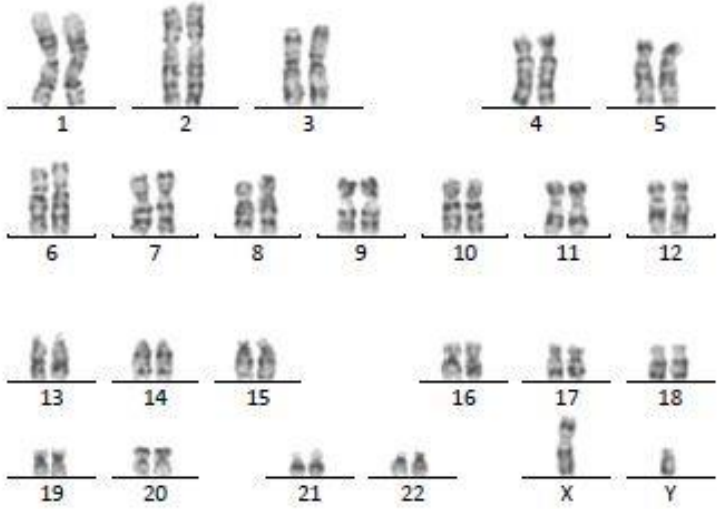
(Genetics Laboratory)

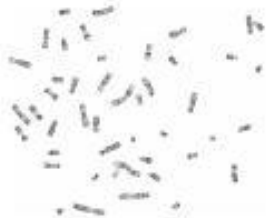
CHROMOSOME ANALYSIS REPORT

Patient ID: CG391B **Patient Name:** Ganesh Patil **Age:** 35 yrs **Gender :** M
Physician Name / Hospital: Dr. S Mane(Paeds)
Specimen: Peripheral Blood
Collected on: 06.04.2021 **Received on:** 06.04.2021 **Reported on:** 14.05.2021
Referral Reason: Parental study taken up because of rob. translocation.



Banding: GTG

Karyotype: 46,XY
Co-ordinates : 94.2/20.1



Impression:
46,XY
Normal Male Karyotype

Note: Analysis is based on the sample received in the laboratory

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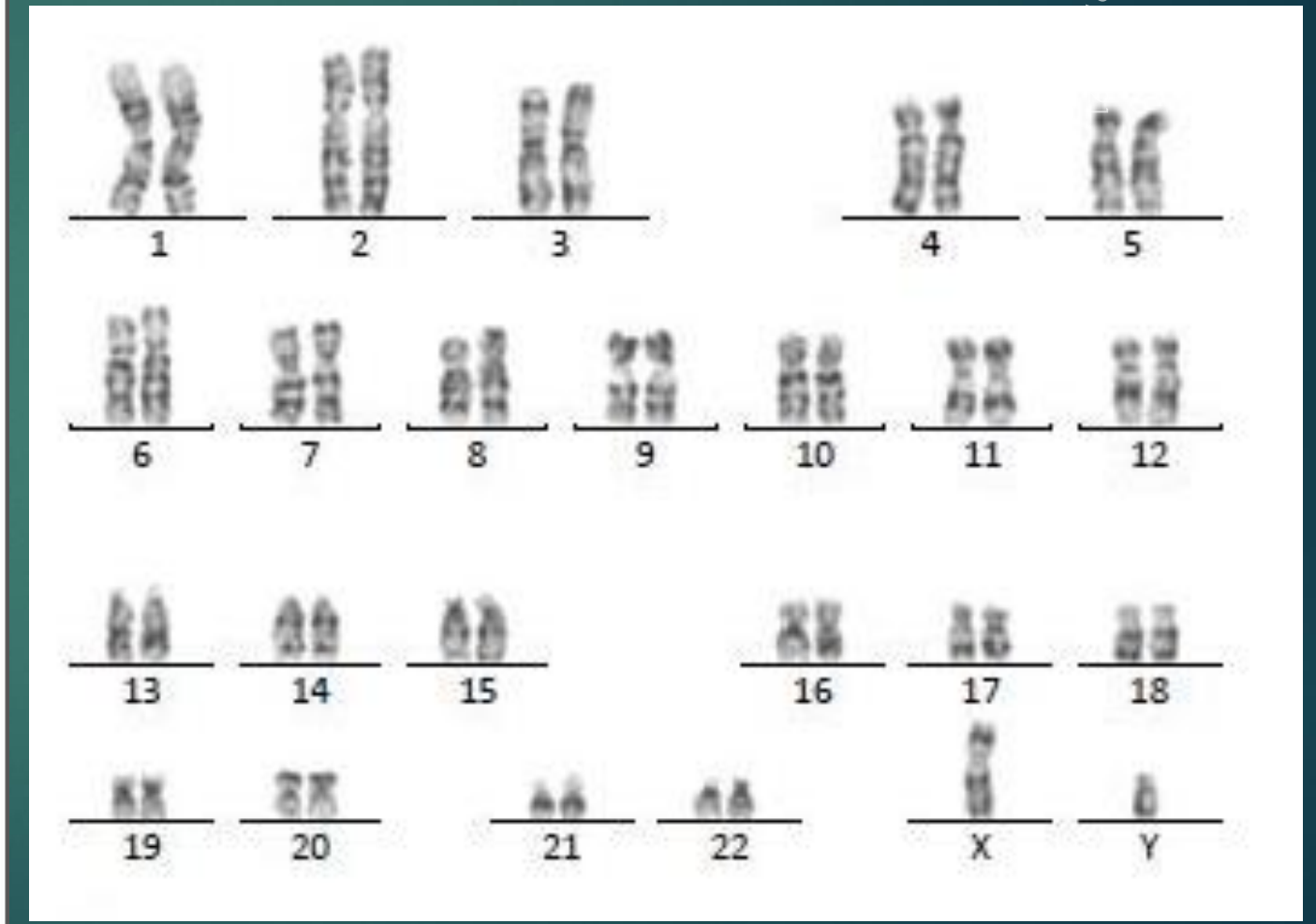
END OF REPORT



MOTHER

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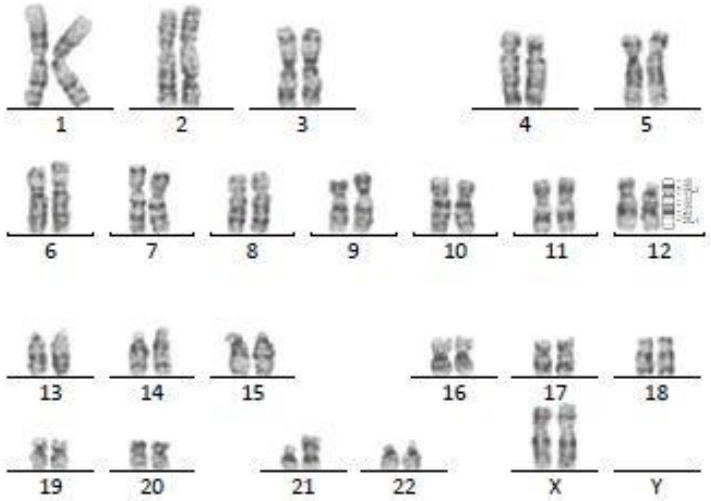
(Genetics Laboratory)

CHROMOSOME ANALYSIS REPORT

Patient ID: CG391A **Patient Name:** Sayali Patil **Age:** 25 yrs **Gender :** F
Physician Name / Hospital: Dr. S Mane (Paeds)
Specimen: Peripheral Blood
Collected on: 06.04.2021 **Received on:** 06.04.2021 **Reported on:** 14.05.2021
Referral Reason: Parental study taken up because of rob. translocation.

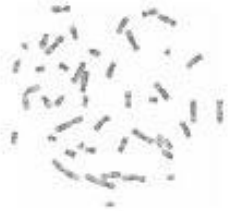


Banding: GTG

Karyotype: 46,XX,+21,rob(21;21)(q10;q10),?del12(p12;pter)
Co-ordinates : 101.3/23.0

Impression:
46,XX,+21,rob(21;21)(q10;q10),?del12(p12;pter)
Note : Deletion 12(p12;pter) needs to be further evaluated by FISH and if necessorry CMA.

Note: Analysis is based on the sample received in the laboratory



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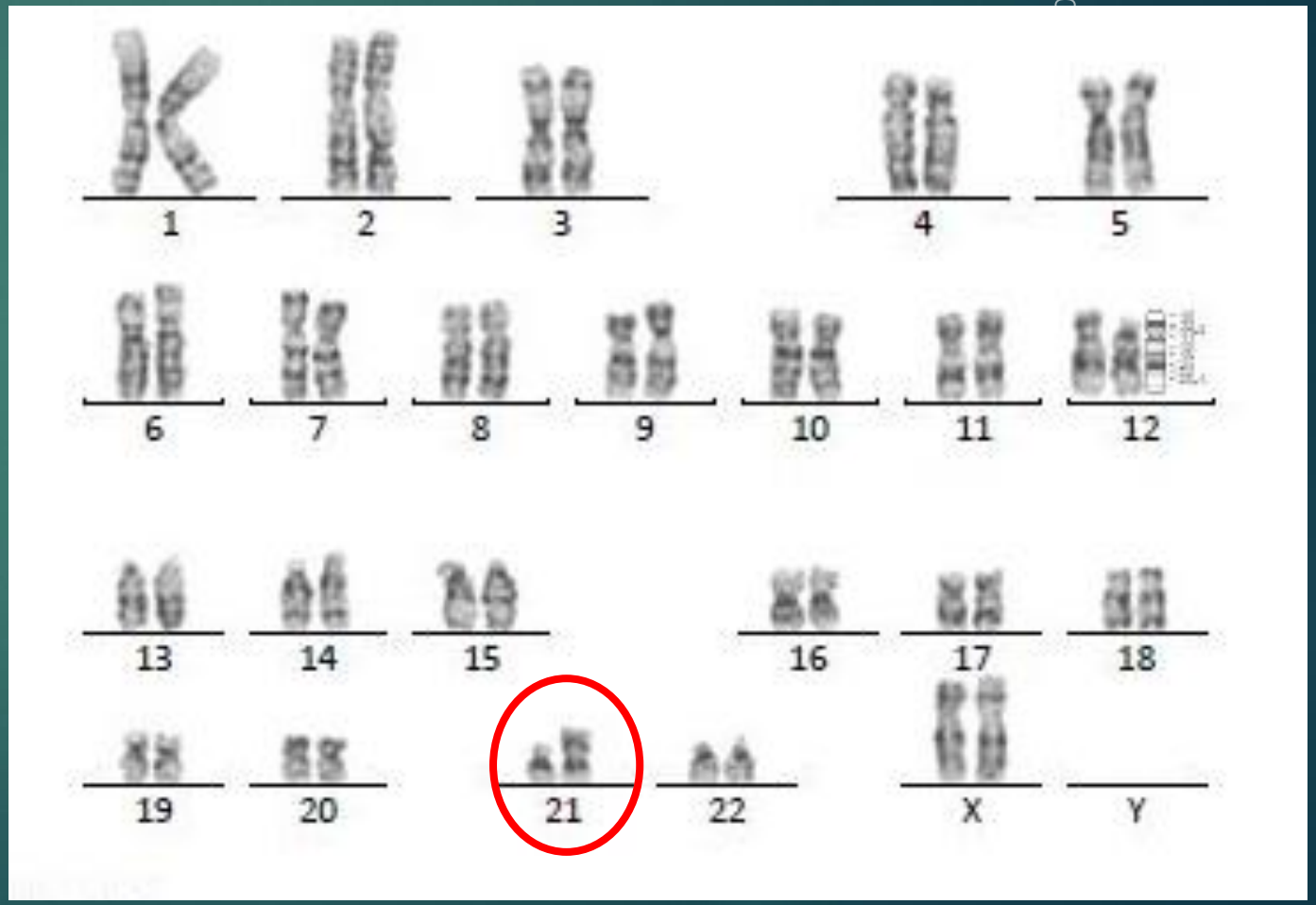


- **Translocation Down syndrorne**
- The importance of Robertsonian translocations is that they can predispose to the birth of babies with Down syndrome as a result of the embryo inheriting two normal number 21 chromosomes (one from each parent) plus a translocation chromosome involving a number 21 chromosome
- The clinical consequences are exactly the same as those seen in pure trisomy 21.
- However, unlike trisomy 21, the parents of a child with translocation Down syndrome have a relatively high risk of having further affected children if one of them carries the rearrangement in a balanced form.

11/25/2021

- Consequently the importance of performing a chromosome analysis in a child with Down syndrome lies not only in confirmation of the diagnosis, but also in identification of those children with a translocation.
- In roughly two-thirds : the translocation occurs *de novo* remaining one-third one of the parents will be a carrier.
- Other relatives might also be carriers.
- Therefore it is regarded as essential that efforts are made to identify all adult translocation carriers in a family so that they can be alerted to possible risks to future offspring.
- This is **TRANSLOCATION TRACING, OR 'CHASING'**.

Risks of recurrence in Robertsonian translocations

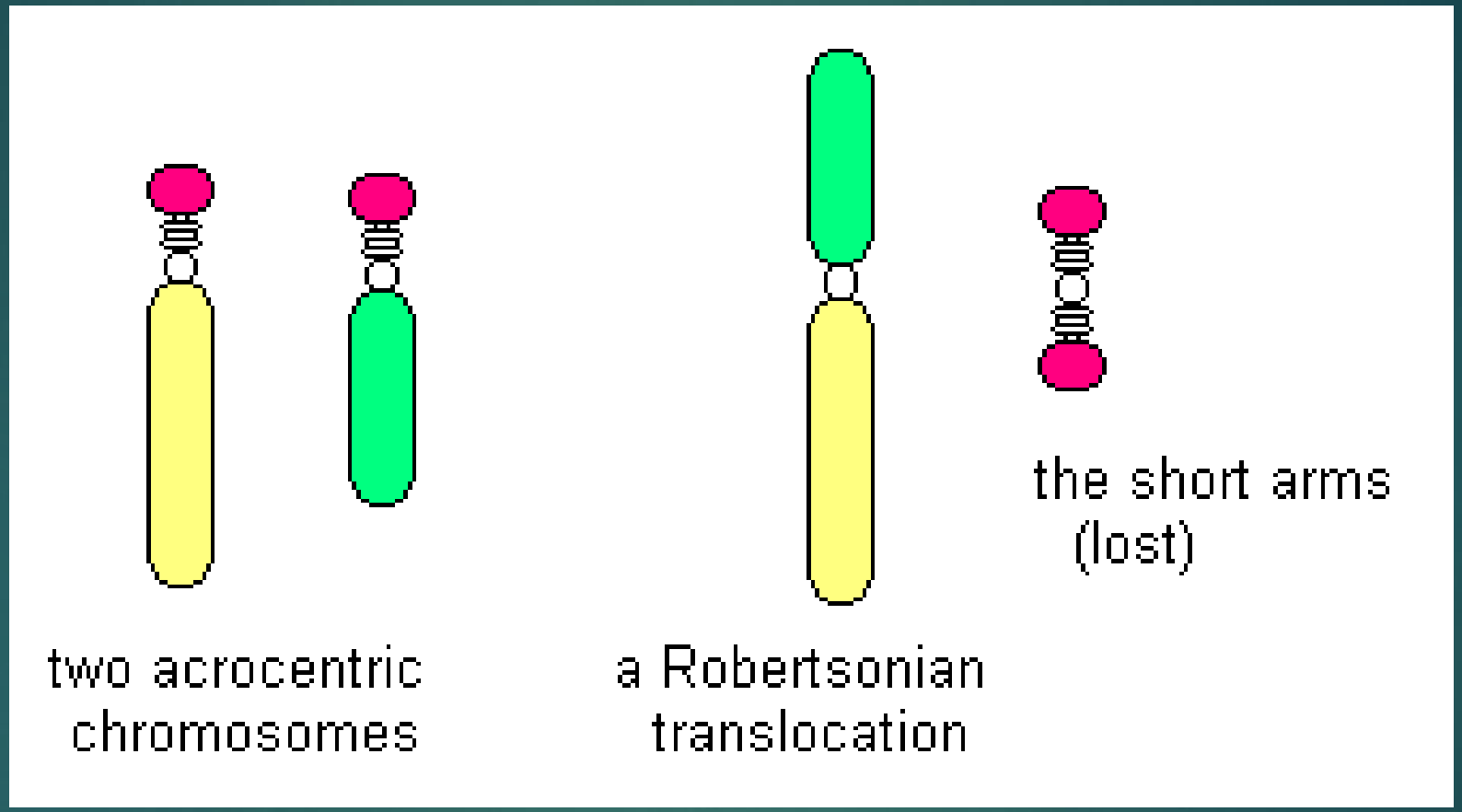
- female carrier of either a 13q21q ora l4q2lqRobertsonian translocation runs a risk of approximately 10 % for having a baby
- male carriers the risk is l-3 %.
- 2lq2lq robertsonian translocation :rare
- All gametes will be either nullisomic or disomic for chromosome

21.

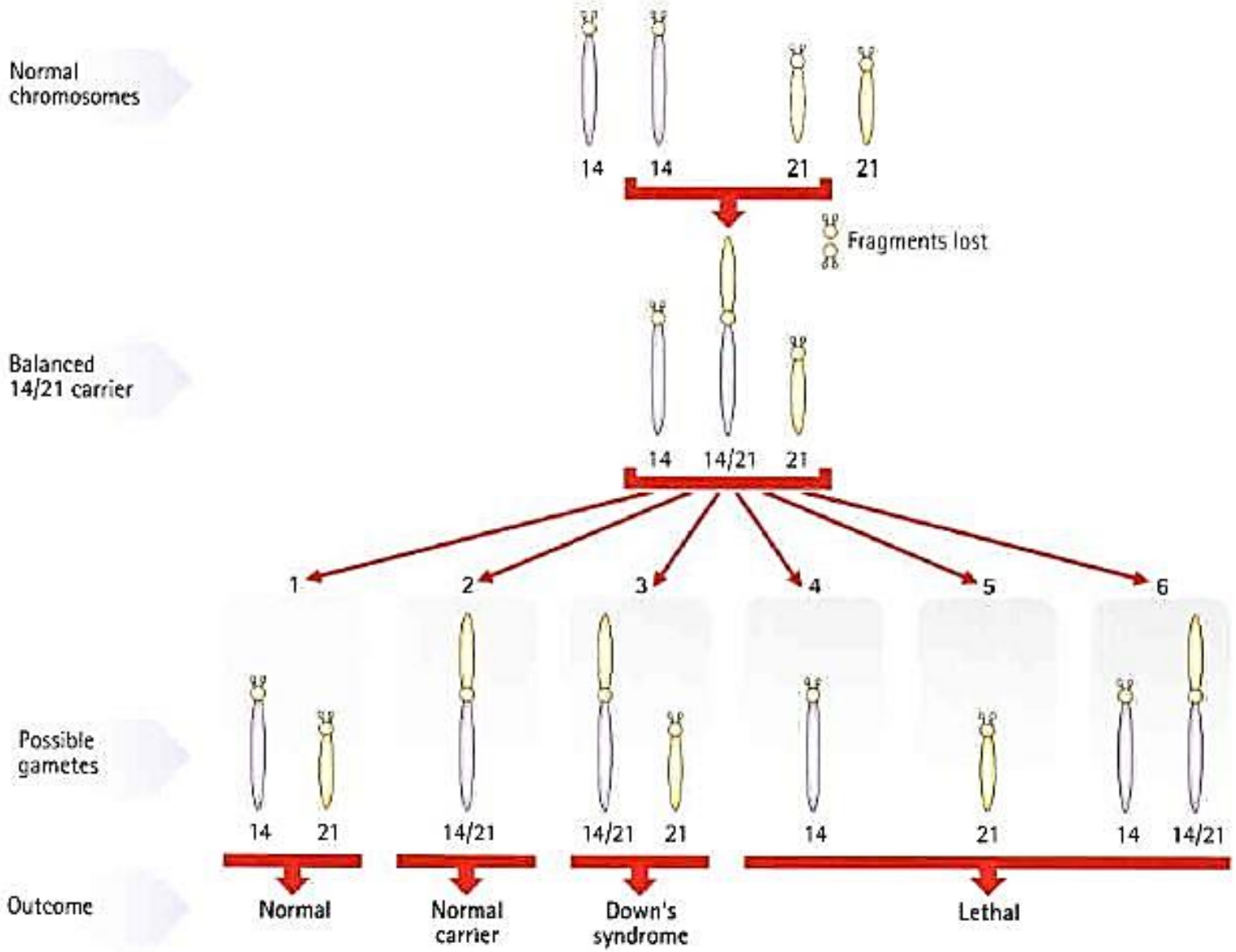
Risks of recurrence in Robertsonian translocations…

- **Consequently all pregnancies will end either in spontaneous miscarriage or in the birth of a child with Down syndrome.**
  Risk of recurrence is 100%

- This is one of the very rare situations in which offspring are at a risk of greater than 50 %of or having an abnormality.



Robertsonian Translocation


Normal chromosomes
14
14
21
21
Fragments lost
Balanced 14/21 carrier
14
14/21
21
1
2
3
4
5
6
Possible gametes
14
21
14/21
14/21
21
14
21
14
14/21
Outcome
Normal
Normal carrier
Down's syndrome
Lethal

MOTHER

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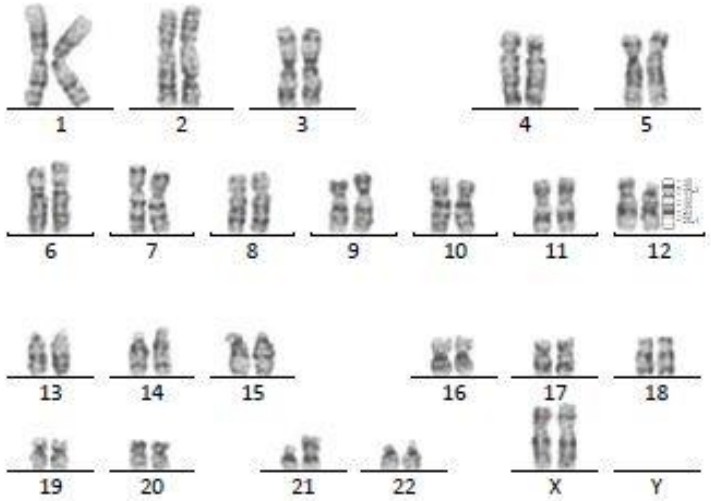

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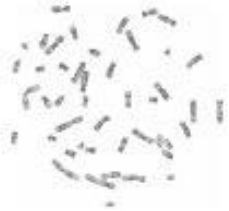

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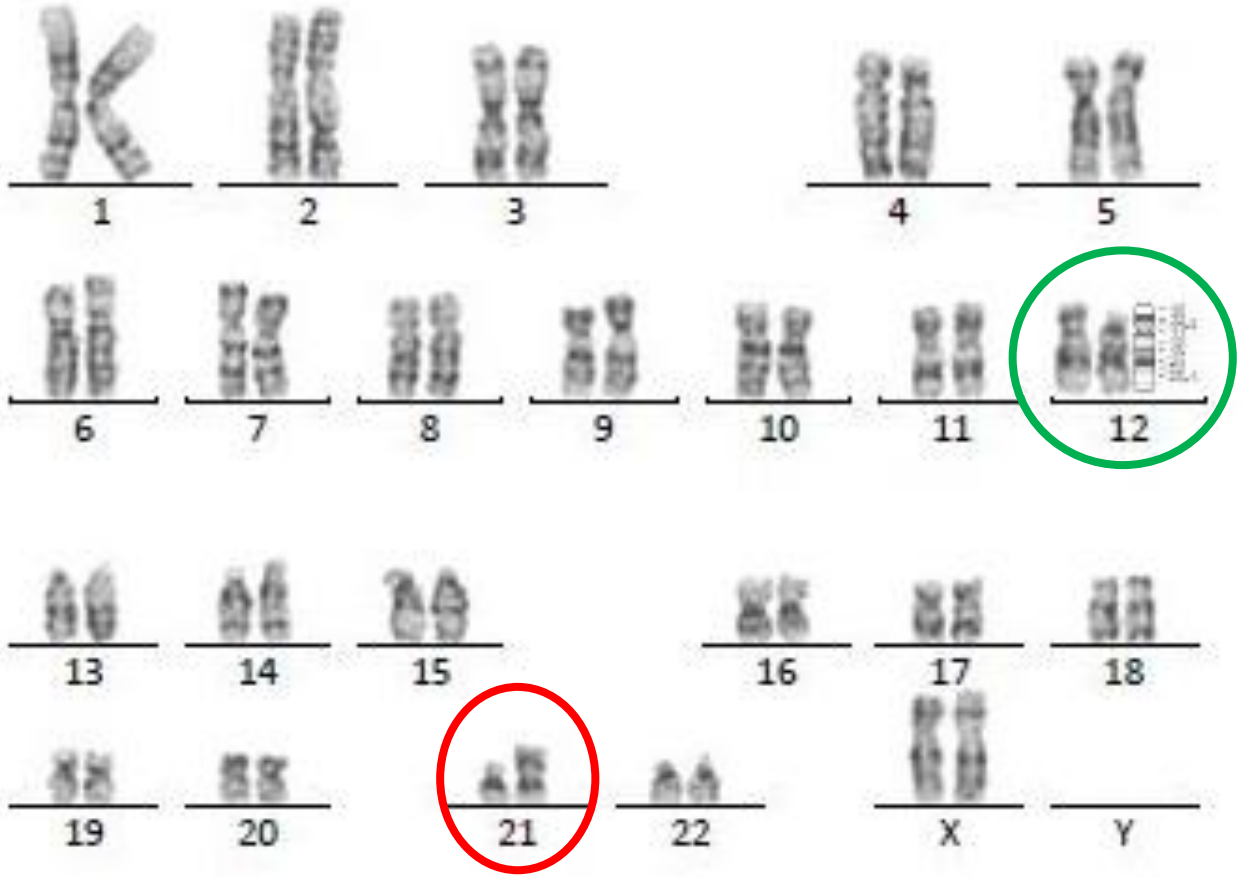

Where did this translocation derive from??

- ? Derivative chromosome formed by the deleted part of chromosome 12 and a translocated chromosome 21!!!!!
- Needs
  - FISH
  - CMA

Cytogenetic Lab Services
Department of Anatomy

5th Floor College Building

Sample collection days:

Monday

Tues day

Friday

Time : up to 1 PM

Send heparinized blood 2ml if prband can't visit lab

Report : Two to three weeks

Detail case workup requested.