



ज्ञान - विज्ञानं विमुक्तये

# Contour Based Segmentation of Chromosomes in G-Band Metaphase Images

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Presented by

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# Study of Birth Defects – NEED OF THE DAY



# Study of Birth Defects – NEED OF THE DAY...

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## World Health Organization (WHO) and March of Dimes (MOD) report on birth defects

- Worldwide -15 million babies are born prematurely and close to 8 million are born with birth defects every year
- Every year an estimated 8 million children--6 percent of total births worldwide--are born with serious birth defects of genetic or partially genetic origin
- 3.3 million children less than 5 years of age die annually and the majority of those who survive may be mentally and physically disabled

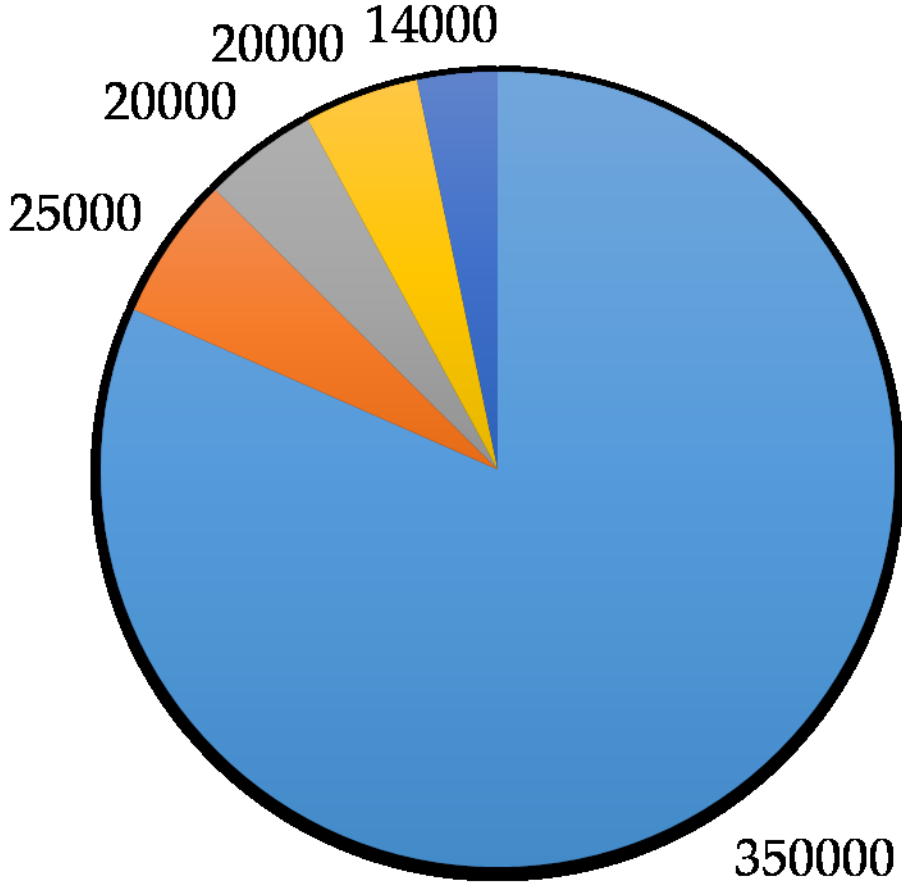


# Study of Birth Defects – NEED OF THE DAY (India)...

Under-five mortality rate (U5MR) (deaths per 1,000 live births)											
1990		2000		2013		Millennium Development Goal target for 2015					
<b>126</b>		<b>91</b>		<b>53</b>		<b>42</b>					
Under-five, infant and neonatal mortality											
Number of under-five deaths (thousands)		Sex-specific under-five mortality rate (deaths per 1,000 live births)		Infant mortality rate (deaths per 1,000 live births)		Number of infant deaths (thousands)		Neonatal mortality rate (deaths per 1,000 live births)		Number of neonatal deaths (thousands)	
1990	2013	1990	2013	1990	2013	1990	2013	1990	2013	1990	2013
<b>3,333</b>	<b>1,340</b>	<b>252</b>	<b>106</b>	<b>88</b>	<b>41</b>	<b>2,339</b>	<b>1,053</b>	<b>51</b>	<b>29</b>	<b>1,362</b>	<b>748</b>



# Origin of the Work



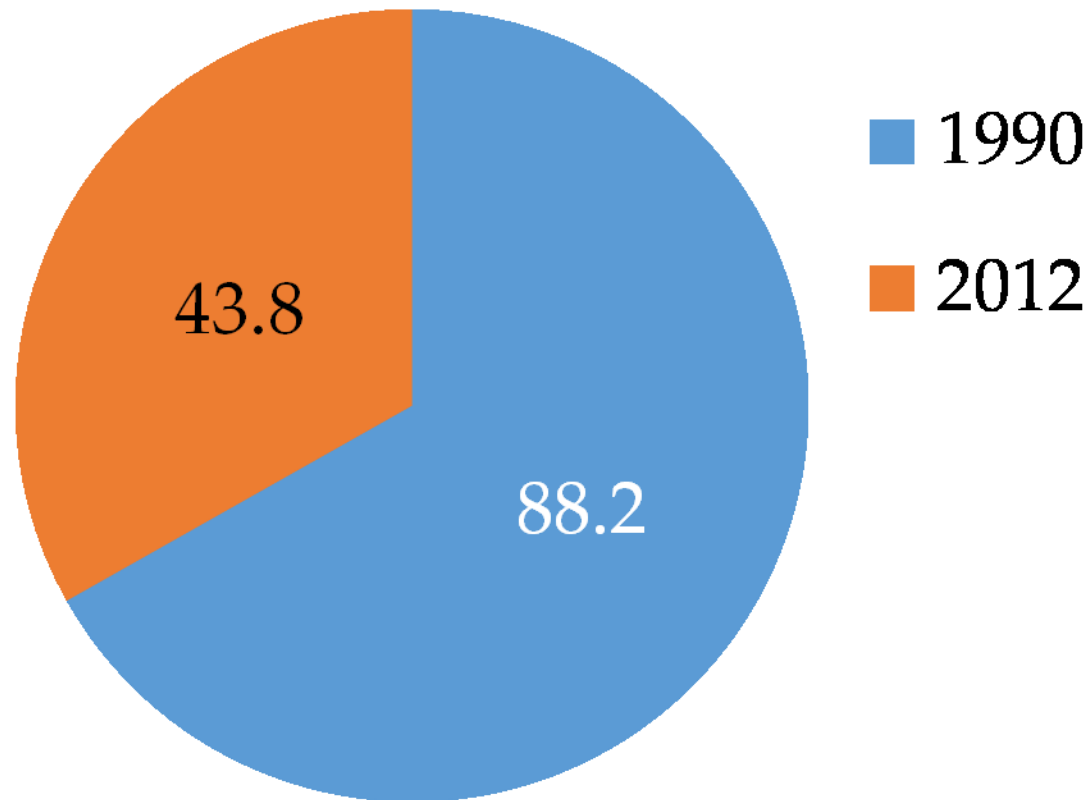
- G6PD Deficiency
- Metabolic Disorders
- Down Syndrome
- Congenital Hypothyroidism
- Thalassaemia

350000 are affected by G6PD Deficiency  
20000 are affected by Thalassaemia  
20000 are affected by Down Syndrome  
14000 are affected by Congenital Hypothyroidism  
25000 are affected by Metabolic Disorders  
100000 are affected by other defects

## Origin of the Work ...

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### OECD Health Statistics on Infant mortality rate 2014 – India



this remains more than ten times  
OECD average  
(deaths per 1000 births)

## Origin of the Work ...

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- India is facing an accelerating demographic switch to non communicable diseases
- New born screening is one of the top 10 public health initiatives in India
- Screening is predictive, preventive, personalized and proactive rather than reactive
- Automated cost effective systems are necessary to reach larger cross section of the population and for effective screening and testing

# Birth Defects

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Abnormality of structure, function or body metabolism present at birth - leading to physical or mental disabilities or death.





# Causes of Birth Defects

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**Genetic** and partially genetic causes, originating mostly before conception (preconception)

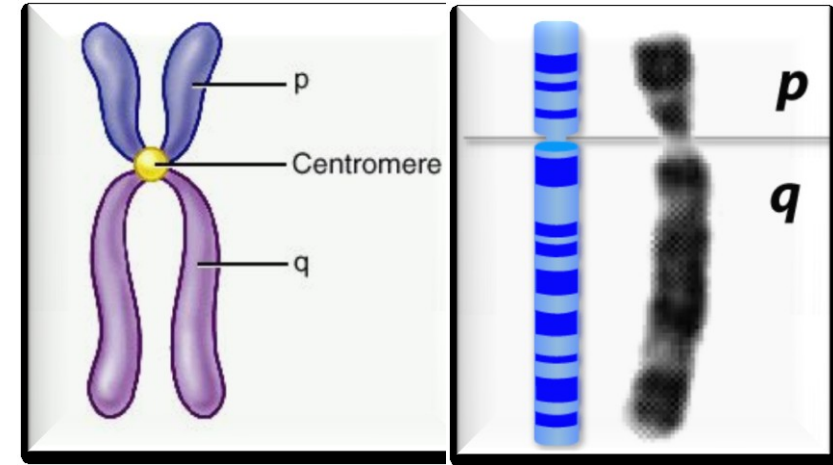
- Most of the birth defects originate prior to conception and are due to abnormalities of chromosomes and genes
- Multifactorial defects occur due to a combination of genes and environmental factors
- Three main categories: **chromosomal abnormalities**, **single gene defects**, **multifactorial disorders**

Causes developing after conception, but before birth (post conception)

– **nongenetic** and caused by intrauterine environmental factor

# Chromosomes

- Humans have 23 pairs of chromosomes (22 pairs are autosomes and the 23rd pair is the sex chromosome referred as X or Y)
- **chromosomal anomaly** – occurs when there is an error in cell division
- **Two types**
  - Numerical anomaly** - variation in number
  - Structural anomaly** - breakage and loss of a portion of chromatid arm or a reunion of arm at different location on the same chromosome or on a different chromosome



# Objective

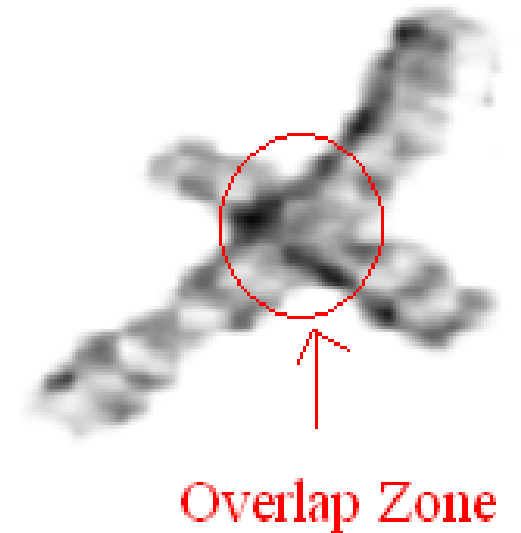
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## ❑ Problem

- Chromosome segmentation becomes extremely difficult when there are more overlapping issues.

## ❑ Solution

- The overlapping chromosome images are segmented by identifying the overlap zone
- The work in the paper is about identifying the overlap zone for performing segmentation



# Methodology

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The algorithm steps followed are

- Image binarization
- Obtaining image contour from binary image
- Obtaining interesting points (concave and convex points) by curvature function
- Identifying overlap zone for finding the number of overlaps in the image.

Overlap zone along with concave points can be used further for segmenting the overlapping chromosomes.

# Methodology....

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The original images taken for the study is shown in Figure 1

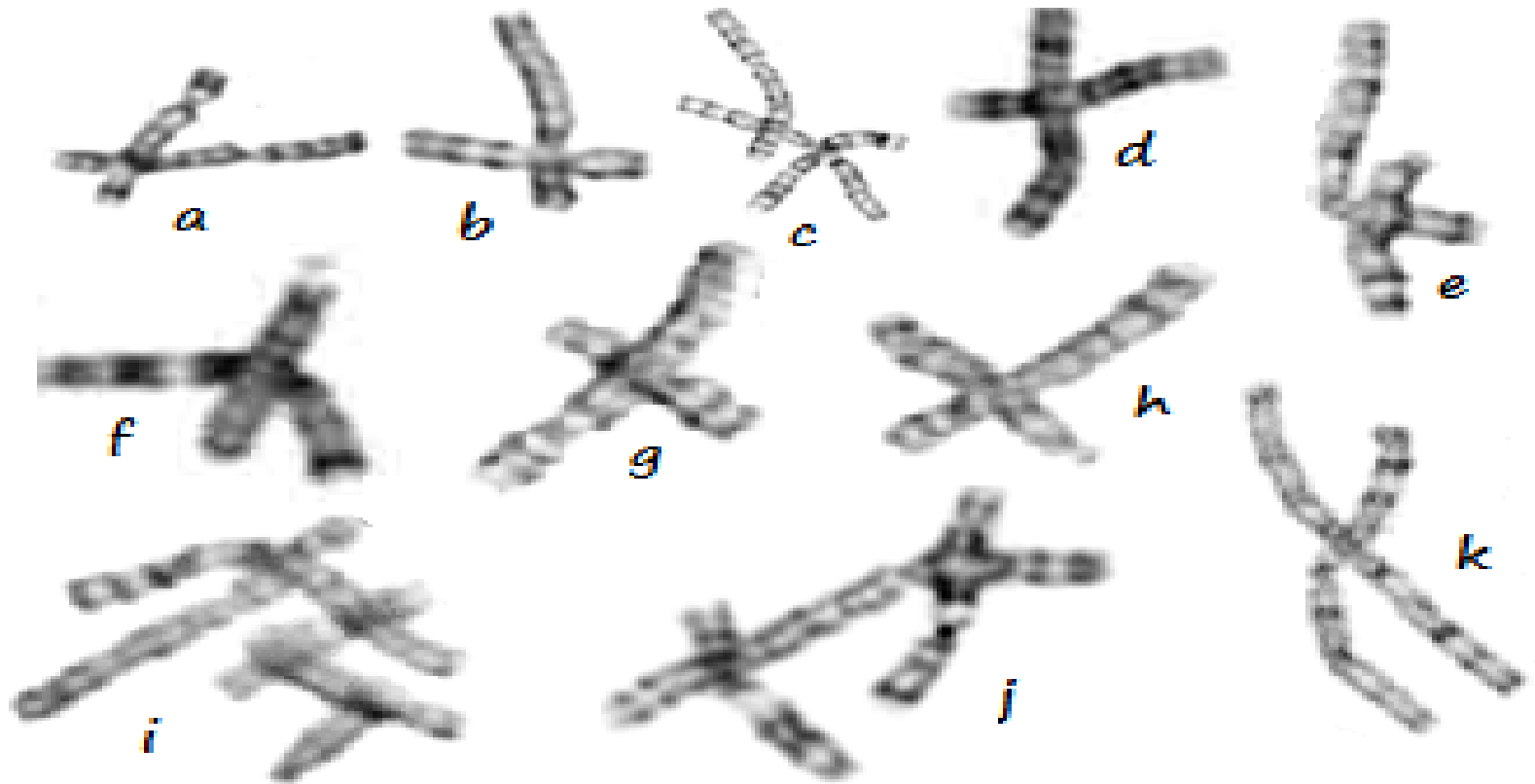


Figure 1 Original Chromosome Images



# Methodology ....

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## Image binarization

- OTSU's thresholding method
  - ❑ Cluster based thresholding method
  - ❑ Converts original image to binary image as shown in Figure 2



Figure 2 Binary Image

## Image Contour

- Helps in determining the shape of the image
- Path between the two pixel points with sequence of distinct pixel coordinates be  $(x_0, y_0), (x_1, y_1), \dots, (x_n, y_n)$  where  $n$  is the length of the path.
- When  $(x_0, y_0) = (x_n, y_n)$  the path is a closed path which is considered as contour as shown in Figure 3



Figure 3 Image Contour

# Methodology....

## Curvature Function

➤ Performed using Parametric Curves

- ❑ First order and second order derivative function is calculated
- ❑ Second order derivative is considered as it give smooth curve

and is represented as

$$C(l) = \frac{x'y'' - y'x''}{(x'^2 + y'^2)^{\frac{3}{2}}}$$

where  $C$  is the point on the curvature function and

$l$  is the curvilinear coordinates.

- ✓ When curve is parabolic – Nature of contour is convex
- ✓ When curve is inverted parabolic - Nature of contour is concave

Figure 4 shows the obtained curvature graph

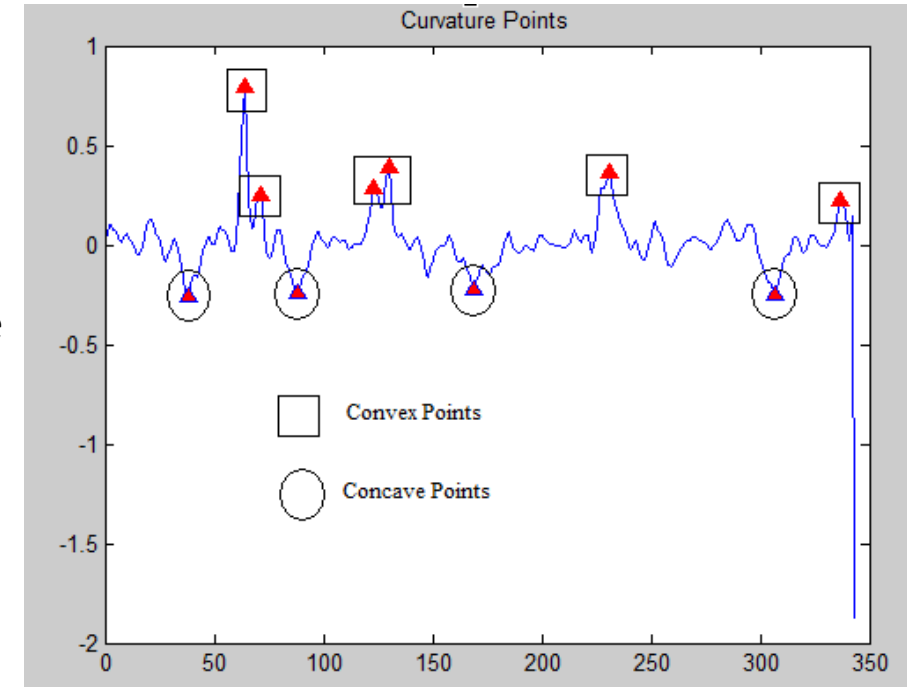


Figure 4 Curvature Graph

# Methodology....

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## Curvature Function

- Helps in locating the concave and convex points on the image
- The obtained concave and convex points are marked on contour and original images as shown in Figure 5a and 5b
- Encircled points in Figure 5a is the required concave points

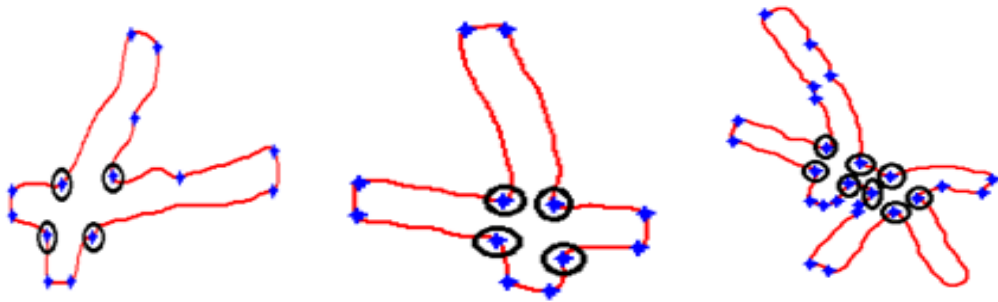


Figure 5a Concave and Convex Points on Contour Image

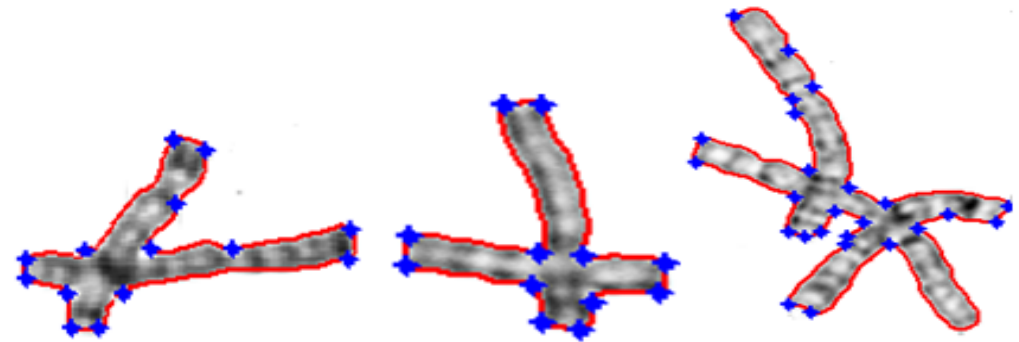


Figure 5b Concave and Convex Points on Original Image

# Methodology....

## Overlap Zone

- Thinning is performed on overlapped image.
- Thinned portion of the overlapped area is considered
- Simple distance transform is applied for obtaining the separation lines of the overlapped region.
- Separation lines with concave points helps in obtaining the overlap zone as in Figure 6a and the zone formation for the input images considered is shown in Figure 6b

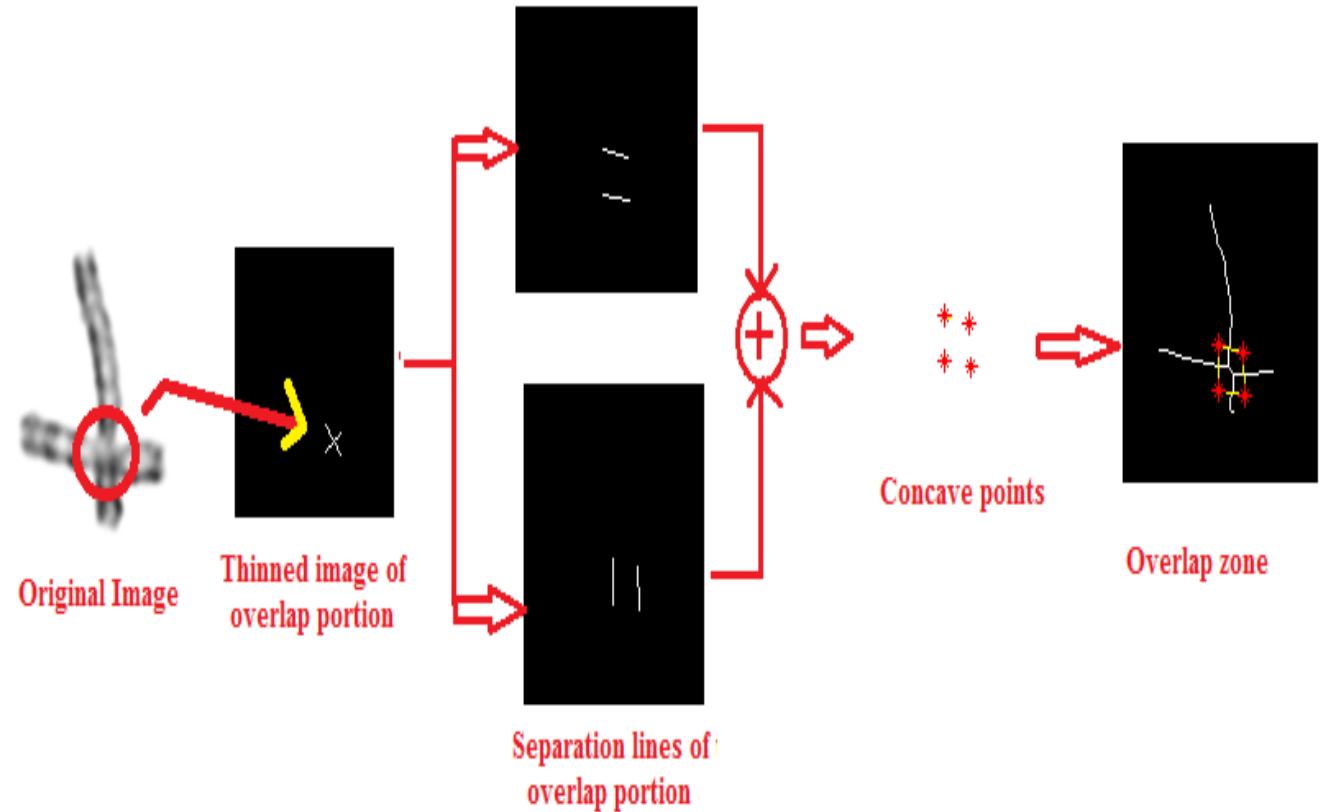


Figure 6a Overlap zone

# Methodology....

Overlap zone for the inputs considered are shown as

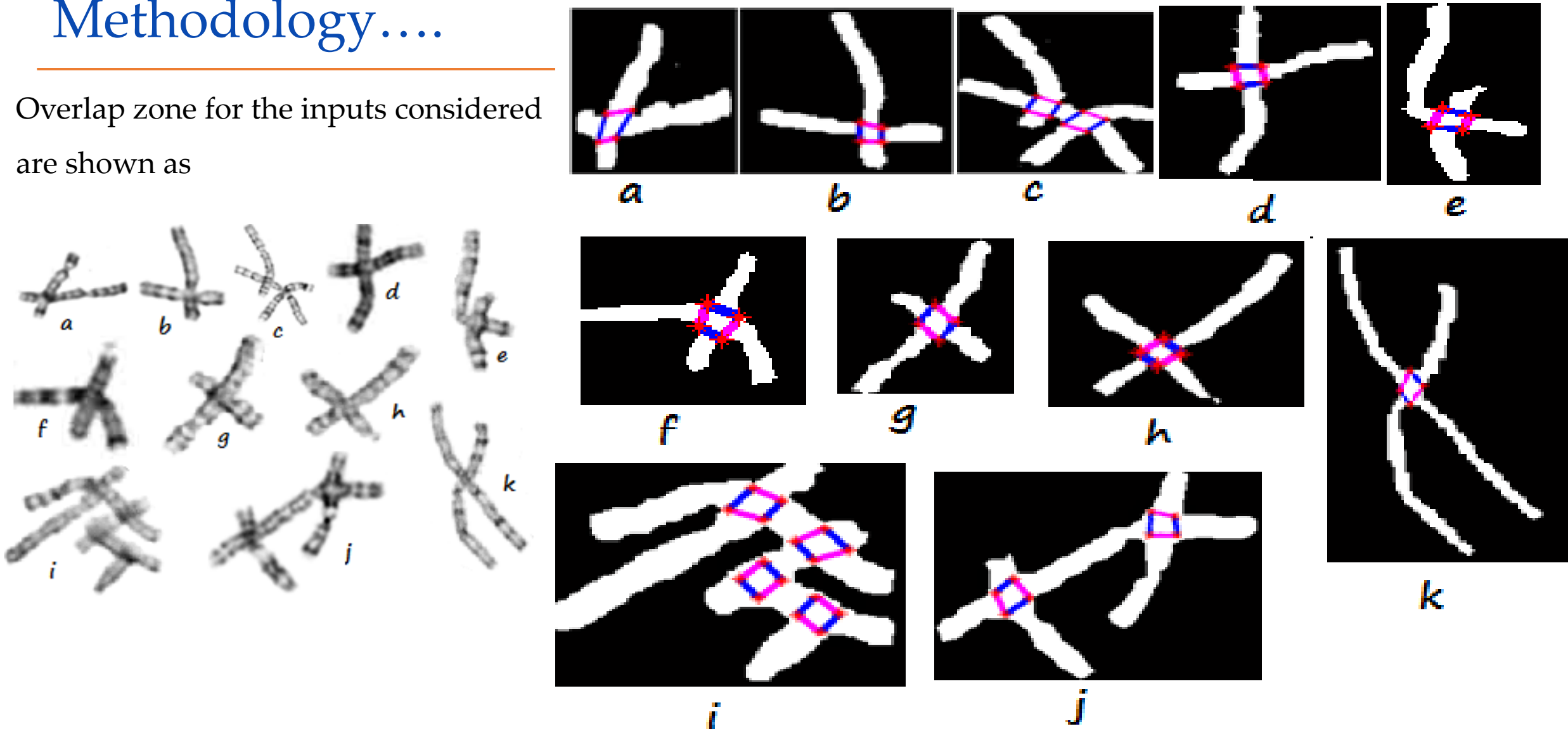


Figure 6b Overlap zone



# Conclusion

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- The algorithm is tested over 350 chromosome images.
- The method works well for identifying the overlapping zone which is the important step in segmentation of overlapping chromosomes with any number of overlaps.
- Future work - Segmentation of overlapped chromosome images can be performed after the identification of the overlap zone.

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- Thank You -